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THE HUMAN GENETIC MUTANT CELL REPOSITORY

List of Genetic Variants, Chromosomal Aberrations and Normal Cell Cultures Submitted to the Repository

Seventh Edition
October 1980

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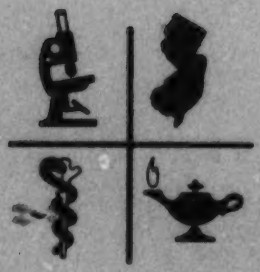

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
Public Health Service
National Institutes of Health

READEX FICHE

HE 20.3452:

THE HUMAN GENETIC MUTANT CELL REPOSITORY

List of Genetic Variants, Chromosomal Aberrations and Normal Cell Cultures Submitted to the Repository

Seventh Edition
October 1980

INSTITUTE FOR MEDICAL RESEARCH
Copewood and Davis Streets
Camden, New Jersey 08103
(609) 966-7377

IMR

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THE NATIONAL INSTITUTE OF GENERAL MEDICAL SCIENCES

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
Public Health Service National Institutes of Health
NIH Publication No. 80-2011
(Revised)

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IN MEMORIUM

During the past year, the staff of the Repository was saddened by the loss of two dedicated scientists whose participation in and enthusiasm for medical genetics will be sorely missed. Dr. William J. Mellman, Professor of Pediatrics and Chairman of the Department of Human Genetics, University of Pennsylvania; and Dr. Robert S. Krooth, Professor and Chairman, Department of Human Genetics and Development, College of Physicians and Surgeons, Columbia University were members of the HGMCR Advisory Committee which guided the early development of the Repository. Both actively participated as submitters, consultants, curators, and senior advisors. The high quality of the cell lines stored in the Repository is in no small measure due to their contributions. Both were pioneers in the use of cultured cells to study the inborn errors of metabolism and they shared their knowledge and experience with all who asked.

We extend our sympathy to the family, friends, and colleagues of both these distinguished men.

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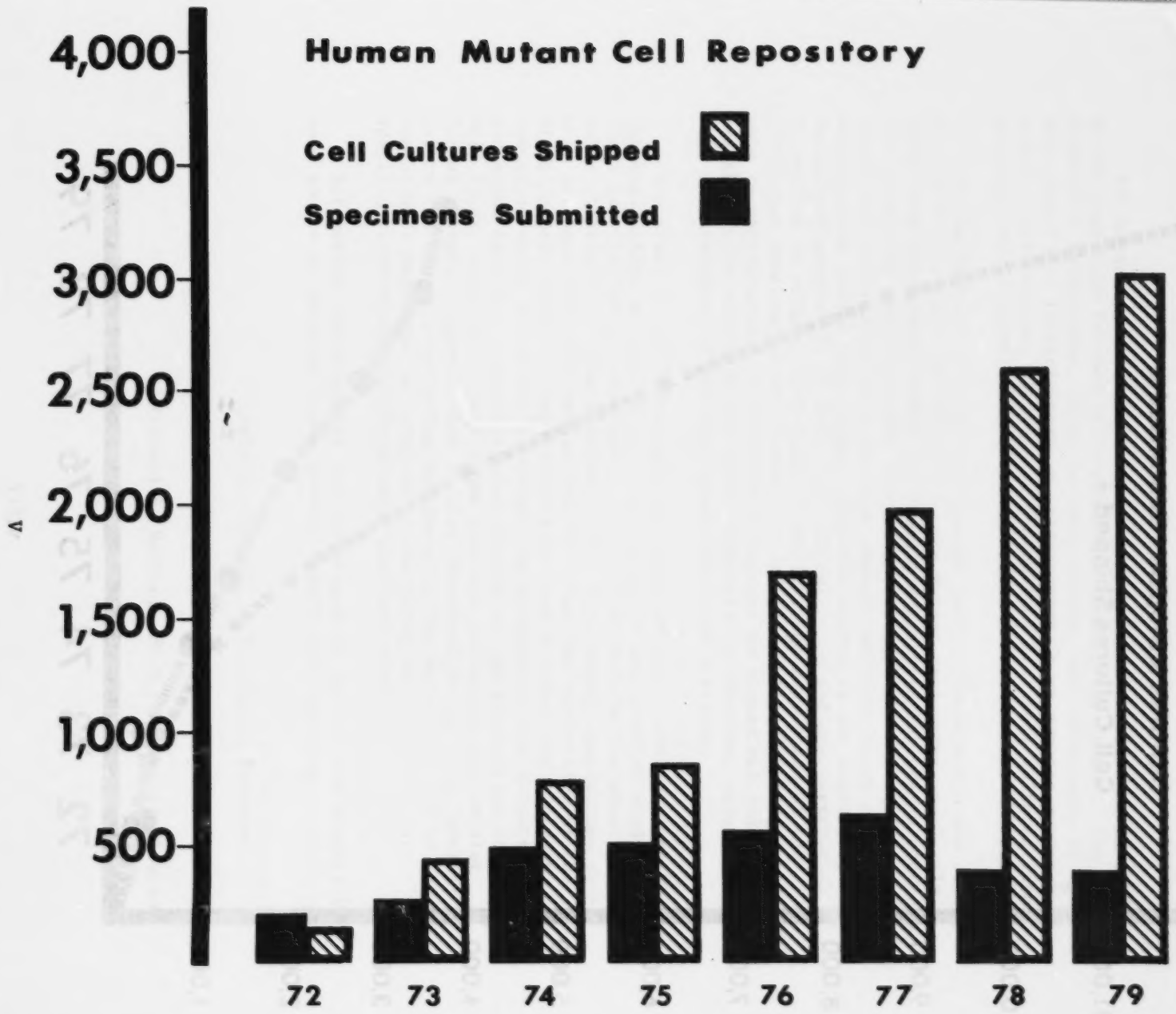
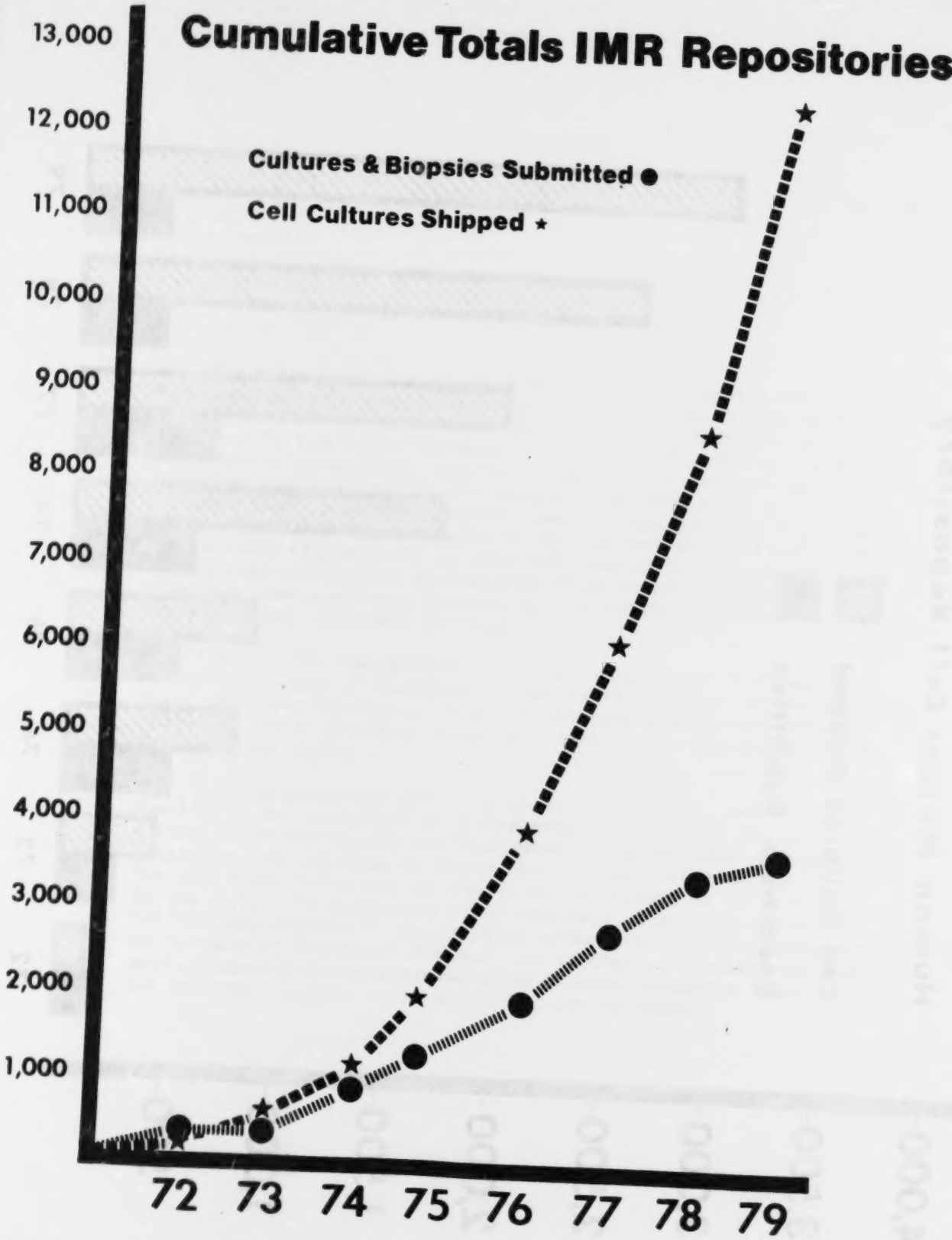
Preface to the Seventh Edition

The seventh edition of the N.I.G.M.S. Human Genetic Mutant Cell Repository catalog contains a number of changes which make it more informative and easier to use. These changes include double spacing between each listing to increase legibility, highlighting the disease diagnosis by underlining, and the replacement of brackets which contained family groups by two columns of numbers which indicate the pedigree (Fam#) to which the individual belongs and from which family member (Famm#) within the pedigree the specimen was obtained. The bibliography (Appendix C) has been enlarged to include approximately one thousand scientific publications which either describe patients from whom specimens were obtained or cite publications in which Repository cultures were utilized.

During 1979, the Repository has continued to be utilized by an increasing number of investigators who desire a source of contaminant free, verified, biochemically mutant or chromosomally aberrant cell cultures. This increased demand is illustrated by the graphs on the following two pages.

The Repository has also continued its efforts to improve the quality of the collection by having experts in various disease catagories either verify the biochemical or chromosomal abnormalities present in the cultures or review the clinical documentation for patients whose mutations are not as yet verifiable in cultured cells. As the collection approaches 4,200 specimens, improving the quality and characterization of the existing collection will take precedence over expanding its size. However, the submission of specimens from well documented patients representing disease catagories which are not currently stored in the Repository are welcomed.


Human Mutant Cell Repository
Cell Cultures Shipped
Specimens Submitted
4,000
3,500
3,000
2,500
2,000
1,500
1,000
500
72
73
74
75
76
77
78
79


Cumulative Totals IMR Repositories
13,000
12,000
11,000
10,000
9,000
8,000
7,000
6,000
5,000
4,000
3,000
2,000
1,000
Cultures & Biopsies Submitted ●
Cell Cultures Shipped ★
72
73
74
75
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79

I. INTRODUCTION

The Human Genetic Mutant Cell Repository was established in 1972 at the Institute for Medical Research in Camden, New Jersey. The Repository establishes, characterizes, stores, and distributes to more than 2,000 investigators in laboratories doing genetic research, highly characterized, viable, and contaminant free cell cultures from patients with biochemical genetic disorders and chromosome aberrations.

The Repository contains low passage cell cultures which are stored in liquid nitrogen. In addition to the mutant cell lines, cell cultures from apparently normal individuals are stored for use as controls. The collection contains human fibroblast, lymphoblast and amniotic fluid cell cultures as well as virus transformed fibroblast cultures and selected animal cell lines. Cultures stored in the Repository are tested and assured to be free from contamination and are verified as to species of origin, karyotype, viability, and expression of the particular biochemical or chromosomal defect. Cell cultures are available to organizations or individuals engaged in biomedical research or health care to assist in the early diagnosis, prevention, understanding, and treatment of the many human inherited diseases.

The staff of the Repository are proud of their contributions to the application of cell culture techniques to the detailed study of human genetics which have lead to a wealth of information about metabolic disorders and gene abnormalities. This highly specialized program has been developed and maintained under the supervision of the NIGMS project officer, many consultants, and a scientific advisory committee which periodically reviews the progress of the Repository and makes specific recommendations related to the acquisition of new cell lines, deletion of cell lines from the collection and catalog format changes. Appreciation is also expressed to the many investigators who submit interesting specimens to the Repository and to those researchers who serve as curators by analyzing cell cultures for specific biochemical and chromosomal abnormalities.

Interested investigators are encouraged to utilize the Repository as a source of verified human genetic mutant cell cultures.

Lewis L. Coriell, M.D., Ph.D., Director

Arthur E. Greene, D.Sc., Associate Director

Richard A. Mulivor, Ph.D., Assistant Director

Staff of the NIGMS Human Genetic Mutant

and NIA Aging Cell Repositories

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Judy Peluse

Microbiology

Judith Sarama, B.S.

Veronica Vanaman, B.A.

Support Services

Al Antanaitis

Thomas Ganor

Harold Ott

Victor Santana

Carole Smith

Robert Wilson

Special Acquisitions

This year approximately 300 new cell cultures were added to the seventh edition of the catalog bringing the total number of listed cultures to 2,080. Of the new listings, the following cultures are of special interest.

GM3569 thru GM3573: Five mouse myeloma cell lines which have been utilized frequently in the production of hybridomas.

GM3542: A fibroblast culture from an individual with Xeroderma pigmentosum; complementation group F.

GM3638 and GM3639: B and T lymphoblastoid cell cultures from an individual with acute lymphocytic leukemia.

GM3402, GM3509, and GM3510: Fibroblast cultures from three individuals with Bloom Syndrome.

GM3534 and GM3540: Lymphoblastoid cultures from two patients with gyrate atrophy of the choroid and retina.

GM3780 thru GM3782: Matched fibroblast and lymphoblast cultures from two brothers affected with Duchenne Muscular Dystrophy.

GM3442, GM3444, GM3446, GM3448, GM3450, and GM3452: Fibroblast cultures derived from skin biopsies of chimpanzees or stumptail monkeys.

II. ORDERING CELL CULTURES

1. REQUIREMENTS

The cells are distributed only to qualified professional persons who are associated with recognized research, medical, educational or industrial organizations engaged in health-related research or health delivery. Before cell cultures can be shipped, the assurance form (Page XIII) must be signed and returned to the Repository. This is adequate for most cell cultures in the Repository. However, lymphoblast or virus transformed cell cultures require prior agreement to observe the Minimum Safety Guidelines (Page XVI) and return of the signed Agreement on Lymphoid and Virus Transformed Cells (Page XV). These forms may be obtained by writing to the Repository, or may be photocopied from the catalog.

2. PROCEDURE

Requests for cell cultures must be submitted on institutional purchase forms including purchase order numbers. Purchasing agents should indicate the name of the investigator on such orders. Telephone orders will be accepted when accompanied by a purchase order number, and should be confirmed within several days by letter. Each culture requested should include the Genetic Mutant Repository number (Rep.#), and the diagnosis.

3. ADDRESS

All requisitions should be addressed to:

Dr. Arthur E. Greene
The Human Genetic Mutant Cell Repository
Institute for Medical Research
Copewood and Davis Streets
Camden, New Jersey 08103

Telephone 609-966-7377

4. FEES

The fee for a 25 cm^2 flask of cells is $30.00, $28.00 each for 2 cultures, $26.00 each for 3 cultures, and $24.00 each for 4 or more different cultures ordered at the same time. Frozen ampules are not shipped, because experience has shown much better success in shipping

freshly recovered growing cultures. Mass production of cell cultures is not a function of the Repository, (See Appendix D, page 218. The Repository is designed to provide only seed cultures. It is suggested that the recipient store aliquots of early passages in liquid nitrogen as insurance against contamination, accidents, artifacts associated with aging, and loss of the culture.

5. SHIPPING CHARGES

The shipping and packaging charges are prepaid and will be added to the invoice at the time of billing.

6. HOW SHIPPED

All cell cultures are grown and frozen in antibiotic-free media to aid in detection and prevention of contamination. Cell cultures in the Repository have been tested and found free of mycoplasma, bacteria, and fungi during characterization, at the time of frozen storage, and after recovery from liquid nitrogen. When an order is received, a frozen ampule is usually recovered from liquid nitrogen on the following Thursday. The culture is inspected for growth and contamination and the medium is changed on Friday. The following Monday, the culture is inspected, the medium is removed and the T25 flask is filled with fresh medium, packed and shipped, usually by air mail special delivery. The flask is enclosed within two watertight plastic envelopes within a styrofoam box in a cardboard mailer to prevent leakage, overheating or freezing during shipment. Return postcards are enclosed. Please return the postcards with notations about the condition on arrival and planned utilization. Pertinent suggestions to improve the effectiveness of the Repository are appreciated. Over the past 5 years 94% of shipments have arrived in satisfactory condition.

7. DESCRIPTIVE DATA

Each shipment contains descriptive data about the cell culture, suggested directions for cell growth, return postcards and pertinent references when available. Upon receipt, cell cultures should be placed in the incubator at 37° C for a few hours or overnight to permit recovery from damage which may have occurred during shipment.

8. REQUEST FOR CITATION AND REPRINTS

It will be greatly appreciated and will make the cell collection more valuable if the source and Repository number are cited in publications in which cell cultures from the Human Genetic Mutant Cell Repository are used. The Repository would appreciate receiving a reprint of such publications.

9. PUBLICATION OF DESCRIPTIVE ARTICLES ON CELL CULTURES STORED IN THE REPOSITORY

Concise descriptions of some of the cultures stored in the Repository are published after characterization in Cytogenetics and Cell Genetics under the name of the individual originally submitting the culture or biopsy. These descriptions include brief clinical histories, assay methods when applicable, family pedigrees, full or partial karyotypes in the case of chromosomal aberrations, and references to previous work involving the cell lines. For a listing of these and other papers, see Appendix C, page 186.

10. ASSURANCE

Before cells can be shipped from the Human Genetic Mutant Cell Repository the recipient institution must submit a purchase order number and agree to the following limitations. This agreement must be renewed annually. Please sign and return originial to the Institute for Medical Research, Copewood Street, Camden, New Jersey 08103.

ASSURANCE FORM

As purchaser of cell cultures from the Human Genetic Mutant Cell Repository we agree that such cells, their progeny, or derivatives will not be used in human experimentation. It is further agreed that if such use is planned, the purchaser will first obtain prior written approval of the Project Officer, NO1-GM9-2101, National Institute of General Medical Sciences.

It is further agreed that cell cultures obtained from the Human Genetic Mutant Cell Repository will not be resold, but they may be replicated by a third party for the original purchaser. The third party shall not be allowed to resell other than to the original purchaser furnishing the material for replication and the third party shall not use such cells for human experimentation.

Name of Institution	Name of Principal Investigator	Date
Signature of Authorized Official	Signature of Principal Investigator	Date

11. AGREEMENT ON LYMPHOID AND VIRUS TRANSFORMED CELLS

This document must be appropriately completed and exchanged before lymphoid and virus transformed cell lines can be transferred from the Human Genetic Mutant Cell Repository to your laboratory.

AGREEMENT

The potential hazardous nature of these human cell lines is unknown. However, in view of this lack of knowledge, it is appropriate that the scientific community be made aware that a potential hazard may exist. Therefore, we recommend that the enclosed guidelines for laboratory procedure be adhered to in the handling of lymphoid and virus transformed cell lines. They are also recommended for handling all cell cultures.

It is understood that you will not further distribute cell lines sent to you to laboratories not under your direct supervision. It is further agreed that all cultures obtained from the Human Genetic Mutant Cell Repository may only be replicated by a third party if the third party also executes an Agreement on Lymphoid and Virus Transformed Cells agreeing to follow the Minimum Safety Guidelines.

As purchaser of cell cultures from the Human Genetic Mutant Cell Repository we agree that such cells, their progeny, or derivatives will not be used in human experimentation. It is further agreed that if such use is planned, the purchaser will first obtain prior written approval of the Project Officer, NO1-GM9-2101, National Institute of General Medical Sciences.

This agreement, when appropriately executed, will be on file and will qualify you to receive lymphoid and virus transformed cell lines from the Human Genetic Mutant Cell Repository.

AGREEMENT ON LYMPHOID AND VIRUS TRANSFORMED CELLS

We the signatories below, have read and understand this document and agree that in the handling of cell lines furnished, we will adhere to the procedures and recommendations outlined in the foregoing and the attached "Minimum Safety Guidelines Recommended for Working with Lymphoid and Virus Transformed Human Cell Lines".

A. For the recipient laboratory:

Investigator: Signature _____ Date_____

Title _____

Head of Laboratory or Immediate Supervisor: Signature _____ Date_____

Title _____

Address _____

B. For Human Genetic Mutant Cell Repository:

Signature _____ Date_____

Title _____

III. HUMAN GENETIC MUTANT CELL REPOSITORY

MINIMUM SAFETY GUIDELINES

RECOMMENDED FOR WORKING WITH LYMPHOID

AND VIRUS TRANSFORMED HUMAN CELL LINES*

A. Supervision

1. Administrative Responsibilities

a. Responsibility of Management

Management should establish a biohazards committee to institute and enforce a health and safety policy which includes a specific safety program for work involving human cell lines. The program should meet applicable federal, state and local regulations and include safety training, maintenance of accident records, and provisions for emergency treatment.

b. Responsibility of the Principal Investigator

The principal investigator is responsible for the preparation of safety protocols for the research program under his direction. The protocols should include appropriate procedures for use, storage, decontamination, disposal and emergency treatment. The protocols should be approved by the biohazards committee and discussed with the research staff before starting the research program.

2. Medical Surveillance and Screening

a. Physical Examinations

Appropriate pre-employment and periodic medical examinations are desirable for persons working with human cell lines.

b. Work Restrictions

Persons having reduced immunologic competency should be restricted from working with these human cell lines.

*These guidelines were developed on February 11, 1974 at a special meeting at the National Institutes of Health attended by representatives from the Office of Biohazards of NCI, NIAID, NIGMS and the Advisory Committee to the Human Genetic Mutant Cell Repository.

c. Serum Collection

Serum should be collected at the time of the pre-employment physical to establish a baseline reference. Serum should be re-collected and stored annually. Serum should be collected immediately after accidental injection or ingestion and at an appropriate interval thereafter. For those individuals exposed to long term lymphoid lines or their derivatives, anti-EB virus titers should be obtained on the collected sera.

3. Laboratory Access

Access to the cell culture area should be restricted to persons directly working with the cell lines, or by specific authorization by the principle investigator or director of the laboratory.

B. Personnel Practices

1. Pipetting

Mechanical pipetting aids rather than mouth pipetting should be used for all pipetting procedures.

2. Eating, Drinking and Smoking

Eating, drinking and smoking should not be allowed in the same areas where cell lines are under study.

3. Protective Clothing

It is recognized that the criteria for protective clothing may vary according to the physical situation of the laboratory and the agents handled. Ideally, adequate protective clothing such as a fully fastened laboratory coat should be worn. This clothing should not be worn outside the work area once the work area has been entered.

C. Physical Control Practices (Recommended for all cell lines, but required for long term lymphoid lines and their deriatives).

1. Ventilated Safety Cabinets or Hoods

Ventilated safety cabinets and hoods and other safety apparatus should be employed and should be tested at least annually to certify correct containment and operation. A list of specifications for satisfactory hoods and instructional materials may be obtained from the Office of Biohazards, NCI.

2. Housekeeping

Appropriate housekeeping procedures which suppress the formation of aerosols should be used. Working surfaces should be wiped down with an appropriate disinfectant before and after work with each cell culture and at the end of the working day.

3. Decontamination and Disposal

Contaminated glassware and similar materials should be appropriately decontaminated or stored for decontamination before removal from the work area for recycling or disposal. Liquid wastes should be decontaminated either chemically or by heat, before being discharged to the community sanitary sewer system.

4. Protection of Vacuum Lines

Vacuum services, if used, should be protected with disposable absolute air filters and liquid traps. The effluent should be collected in liquid traps containing concentrated disinfectant.

References

Biohazards in Biological Research, ed. A. Hellman, M.N. Oxman and P. Pollack. Cold Spring Harbor Laboratory, New York (1973).

National Cancer Institute Specification: General Purpose Clean Air Biological Safety Cabinet.

IV. SUBMISSION OF SPECIMENS TO THE REPOSITORY

Biopsies are preferred to established cell cultures because they can be processed and stored in lower passage. When submitting a specimen to the Repository, the biopsy should be placed in a 25 cm^2 tissue culture flask or screw top vial with culture medium containing 100 units/ml of penicillin and 100 mcg/ml of streptomycin. Tape the top or cap of the flask very securely to prevent leakage. Package the biopsy flask in a container so that it will not be broken in transit. Mark on the outside of the package that it should be kept at room temperature and not refrigerated, frozen or overheated.

Early passage cultures may be submitted when a biopsy cannot be obtained from the patient. The culture flask (25 cm^2 preferably) should be filled to the top with culture medium and shipped as described above. Mail biopsy or culture prepaid by air mail special delivery to:

Dr. Arthur E. Greene

Human Genetic Mutant Cell Repository

Institute for Medical Research

Copewood and Davis Streets

Camden, New Jersey 08103

Phone 609-966-7377

Before a biopsy or cell culture can be processed for the Repository, clinical and laboratory documentation proof of the diagnosis must be provided on a submission sheet which is available from the Institute for Medical Research, or may be photocopied by the submitter from pages XX-XXIII of this catalog. Without this information the culture cannot be coded into the information retrieval system or be of value to users of the Repository. To conserve effort and expense, the Repository staff are instructed not to process a specimen unless documentation of diagnosis is provided with the specimen. Submitters are therefore requested to fill in all applicable blanks on the submission sheets and provide a family pedigree if available. If a chromosomally aberrant specimen is submitted, inclusion of a photograph of the karyotype is appreciated.

No biopsies or cell cultures submitted to the Human Mutant Cell Repository are to be obtained from a live fetus, defined by the presence of pulse, circulation and other vital signs.

TO BE FILLED IN BY

Repository Number _____
Date Received ___/___/___
Mo. Day Y

SUBMISSION SHEET

Please Complete Every Item that Applies

Submitting Investigator _____

PATIENT INFORMATION:

SPECIES: HUMAN _____ OTHER _____

PATIENT'S INITIALS: _____

AGE: _____ (circle one)
NB (newborn) DA (days) WK (week
MO (months) YR (years)
If Fetal Tissue _____ FW (weeks of gestati

PHENOTYPIC SEX: (circle one) M (male) F (female) A (ambiguo

RACE: (circle one) BL (black) WH (white) AI (American Indi
OR(oriental) IN (Asiatic Indian) Other _____

ETHNIC BACKGROUND (if relevant): _____

CLINICAL PHENOTYPE: A (clinically affected) C (clinically norm
H (heterozygous) R (at risk) V (variant) O (oth
B (balanced chromosome variant) U (unbalanced chromosome varia

DIAGNOSIS: _____

McKUSICK NUMBER _____

CLINICAL MANIFESTATIONS: (Please include detailed clinical reports wh
available) _____

_____

_____

SOURCE OF CLINICAL INFORMATION:

____ 1. Personal Examination
____ 2. Hospital Records
____ 3. Genetic Clinic Records
____ 4. Specialist's Report
____ 5. Autopsy Records
____ 6. Private Physician
____ 7. Other _____

IS A PEDIGREE INCLUDED: (circle one) Y (yes) N (no)
(Please include a detailed pedigree if it is available)

LABORATORY INFORMATION: Local I.D. Number _____

TYPE OF SAMPLE SUBMITTED: (circle one)

Culture
- C (fibroblast culture)
- L (lymphocyte culture)
- A (amniotic fluid cell culture)
- E (differentiated cell culture)
- Other _____

Tissue Sample
- B (skin biopsy)
- P (peripheral blood)
- Other _____

Passages of culture submitted _____

Biopsy Site _____

Population doubling level _____

Date culture was established _____

CULTURE MEDIA in which submitted:

McCOYS 5A	_____ %	HAMS F-10	_____ %
HANKS-EAGLES	_____ %	RPMI 1640	_____ %
EAGLES-EARLES	_____ %	OTHER	_____
HAMS F-12	_____ %	FETAL BOVINE SERUM	_____ %
		Inactivated	_____ I
		Uninactivated	_____ U

ADDITIVES ___Y___ (yes) ___N___ (no)

ANTIBACTERIAL
- ____ Penicillin
- ____ Streptomycin
- ____ Gentamycin
- ____ Other

ANTIFUNGAL
- ____ Fungizone
- ____ Mycostatin
- ____ Other

REQUIRED NUTRIENT
Specify _____

Special Instruction on growth, handling, or freezing: _____

ADDITIONAL PERTINENT INFORMATION:

Has this patient/family been reported in the literature: Y (yes) N (no)

Please cite reference(s): _____

For the disease category you are submitting, is the defect expressed in

Fibroblasts Y (yes) N (no)
Lymphoblasts Y (yes) N (no)

DIAGNOSTIC LABORATORY TESTS AND STUDIES

BIOCHEMICAL STUDIES: Y (yes) N (no)

Were the studies done on this culture? Y (yes) N (no)

If no, what tissue or culture? _____

TYPE OF ASSAY _____

_____

METHOD USED _____

_____ (please cite referen

RESULTS FOR PATIENT? _____

RESULTS FOR CONTROLS? _____

RESULTS ARE: (circle one)

1. Consistent with Homozygosity
2. Consistent with Heterozygos
3. Inconclusive
4. Normal levels
5. Other _____

CHROMOSOME STUDIES: Y (yes) N (no)

Where the studies done on this culture? Y (yes) N (no)

If no, what tissue or culture? _____

Current Paris Nomenclature _____

_____

The Chromosome variant is: B (balanced) U (unbalanced)

Staining methods used: _____

Other Chromosome analysis: _____

_____ increased breakage? _____ sister chromatid exchang

Have other specimens from this patient/family been stored in the Repository in Camden? Y (yes) N (no)

If yes, what are their Repository numbers and what are their relation-

ships: _____

_____

ADDITIONAL REMARKS:

_____

_____

_____

_____

RELEASE, PERMISSION AND CONSENT FORM

To encourage storage of unusual cell cultures in the Repository, provision has been made for delayed release to other investigators if the contributor so desires. Please check your preference: a) release culture to anyone requesting it _____, b) release only to contributor during the first year _____. At the conclusion of one year the cell culture will be listed in the next printing of the catalog and made available to other investigators unless the submitter obtains a delay from the project officer at N.I.G.M.S.

I hereby grant permission for these cells to be stored in a bank of genetic mutant cell cultures and the progeny cells distributed to qualified investigators. Appropriate consent was obtained from the patient from whom the cells were originally obtained, or can be reasonably inferred, for use of these cells for diagnosis, research, teaching or therapy.

No biopsies or cell cultures submitted to the Human Mutant Cell Repository are to be obtained from a live fetus, defined by the presence of pulse, circulation and other vital signs.

Date _____ Submitter _____
(Signature)

Address _____

_____

Telephone Number _____

Mail completed form with, or preferably preceeding shipment of cell cultures, to: Dr. Arthur Greene, Institute for Medical Research, Copewood and Davis Streets, Camden, New Jersey 08103.

Telephone (609) 966-7377.

Use of the Catalog

A. Locating a Cell Culture

The overall format of the catalog can be seen by consulting the Table of Contents.

A cell culture belonging to a particular disease category may be found by consulting the alphabetical index of diagnoses comprising Appendix F, page 244. Following each diagnosis is the page number on which that disease may be found. The letters preceding the page number indicate the section of the catalog to which that page belongs: F, fibroblast; L, lymphocyte; A, amniotic; SV, virus transformed; AN, animal; and AG, aging.

If the Repository number of a culture is known, its page location may be identified by consulting Appendix E, page 220 which lists Repository number versus page number. Fibroblast, Lymphoblast, and Amniotic Fluid cell cultures from the same individuals or family groups are cross referenced in the remarks column for each cell line.

B. Explanation of the Data Columns of the Catalog

COLUMN

REP #: The Repository number of the cell culture is the number assigned to it upon receipt at the Human Genetic Mutant Cell Repository. It consists of two letters and four digits. i.e., GM1234. Fibroblast cultures originate from skin unless otherwise noted. Lymphocyte cultures usually originate from peripheral blood.

Passage #: The number of serial in vitro transfers of the cell culture as stored in liquid nitrogen.

Culture Medium: For explanation of the code see Appendix A, page 159.

Submitter Code: A number which identifies the investigator who submitted the specimen to the Repository. For code see Appendix B, page 160.

Age: The age of the donor is expressed in years or, when appropriate, in months (MO), weeks (WK), days (DA), or fetal weeks of gestation (FW).

Sex: Male (M), Female (F), or Ambiguous (A)

Race: WH-Caucasian; BL-Black; OR-Oriental; IN-Indian (India); AI-American Indian; BR-Polynesian. Space left empty if race unknown.

Genetic Status:

+	Normal gene
-	Mutant gene
y-	Hemizygous for X-linked trait
+?	Genetic status not determined
+-(0)	Carrier for trait as determined by pedigree or clinical diagnosis

The McKusick* number immediately following the name of the disease indicates the dominant, recessive, or X-linked nature of the disease. The first digit is one for a dominant disease (e.g. Huntington Chorea - 14310), two for a recessive disease (e.g. Tay-Sachs Disease - 27280), and three for an X-linked trait (e.g. Lesch Nyhan Syndrome - 30800).

Verified: The clinical diagnoses of all cell cultures listed in the catalog have been verified by the submitter and reviewed by the staff and one or more members of the Advisory Committee. B is entered in this column if the defect was verified on the cell culture before freezing; A is entered if verified after recovery from liquid nitrogen. The space is left blank if the defect was verified on another cell culture or tissues from the same patient or relative, or if verification is not possible because the defect is unknown or not expressed in culture.

Paris Nomenclature: As described (Paris Conference 1971) and Supplement (1975): Standardization in Human Cytogenetics. Birth Defects: Original Article Series, VIII:7, 1972, and XI: 9, 1975. The National Foundation, New York.

Balanced, Unbalanced: Balanced (B) or Unbalanced (U) karyotype

Relation to Proband: Example: AFF BRO = Affected brother.

Fam #: A unique pedigree number is assigned if cultures from more than one member of a family or more than one sample type (i.e. fibroblast and lymphoblast) from a single individual are stored in the Repository.

Famm #: Within each pedigree, each family member is assigned a unique family member number. Cultures having the same Famm# were obtained from the same individual.

* McKusick, Victor. Mendelian Inheritance in Man 5th Edition. Johns Hopkins Press, Baltimore, London. 1978.

Remarks: Includes any pertinent information that is not found in the preceding columns and cross references to other samples from the same individual and/or other members of the same family.

* Before a Repository number indicates that a literature reference for this cell line is found in Appendix C, page 186.

NIA Cell Culture Repository

The National Institute on Aging (NIA) established in 1974 a human cell culture repository at the Institute for Medical Research, Camden, New Jersey. This was to facilitate cellular aging research programs and to contribute to the elucidation of mechanisms in human aging at the cellular and molecular level. This collection contains cell lines from individuals who express aging associated degenerative processes including premature aging syndromes, diseases with known or suspected DNA repair dysfunctions, neurological diseases associated with aging and conditions which apparently predispose to altered cellular proliferative capacity in vitro. There are a variety of cell cultures from well documented "cancer families" as well as several lines derived from apparently normal cells transformed by SV40 virus infection. The Aging Cell Culture Repository also includes a number of apparently normal fibroblast cultures from fetal and non-fetal origin. This group includes the extensively banked diploid fetal lung fibroblast cultures, IMR-90 (human female) and IMR-91 (human male) which are available at various population doubling levels.

The workscope of the NIA Cell Culture Repository also includes cytogenetic characterization of cell lines for NIA grantees or contractors who do not have such a facility at their disposal.

The Repository staff is interested in obtaining biopsies or cultures from well documented patients and family members in the categories listed. We also wish to include a variety of differentiated cell types where technology permits liquid nitrogen storage and cell culture distribution. The Aging Cell Culture Repository is supported by the National Institute on Aging contract #NO1-AG-0-2100.

Requests for cells and information may be addressed to:

Dr. Arthur E. Greene
Institute for Medical Research
Copewood Street
Camden, New Jersey 08103
Telephone 609-966-7377

APPARENTLY NORMAL HUMAN FIBROBLAST CELL CULTURES

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
Apparently Normal: Fetal Tissue										
GM0468	2	MC5A	20U 0026	FW	M		PROBAND			46,XY; 14.5 cm crown-rump length; skin biopsy
*GM0011	2	MC5A	20U 0026	8FW	M	BL	PROBAND			46,XY; skin biopsy
*GM0009	3	MC5A	20U 0026	12FW	M	WH	PROBAND			46,XY; skin biopsy
*GM0010	2	MC5A	20U 0026	12FW	M	WH	PROBAND	166	1	46,XY; skin biopsy; 12 cm crown-rump length; same patient as GM1380
GM1379	4	EEMEM	20U 0026	12FW	M		PROBAND	167	1	46,XY; lung biopsy; 10.5 cm crown-rump length; same patient as GM1381
GM1380	13	EEMEM	20U 0026	12FW	M	WH	PROBAND	166	1	46,XY; lung biopsy; 12 cm crown-rump length; same patient as GM0010
*GM1381	4	EEMEM	20U 0026	12FW	M		PROBAND	167	1	46,XY; skin biopsy, 10.5 cm crown-rump length; same patient as GM1379
GM1603	2	MC5A	20U 0152	12FW	M	BL	PROBAND	168	1	46,XY; skin biopsy; same patient as GM1604
GM1604	2	MC5A	20U 0152	12FW	M	BL	PROBAND	168	1	46,XY; lung biopsy; same patient as GM1603
Apparently Normal: Non-Fetal Tissue										
*GM0970	4	EEMEM	20U 0095	3DA	M	WH	PROBAND			Skin biopsy
*GM3468	8	HF12	20U 0331	5DA	M	WH	PROBAND			Foreskin; sensitive to azaguanine & thioguanine; utilizes exogenous hypoxanthine; HPRT normal
GM2936A	3	HF12	20U 0185	20DA	M	WH	PROBAND			Mexican; skin biopsy
*GM0041	5	MC5A	20U 0045	3MO	F	WH	DAU	162	1	46,XX; Puerto Rican; skin biopsy; mother is GM0037A
*GM0302	6	MC5A	20U 0095	10MO	M		PROBAND	331	1	Skin biopsy; formerly GM0075; normal B-hexosaminidase and B-galactosidase
*GM0969	4	EEMEM	20U 0095	2	F	WH	PROBAND			46,XX; skin biopsy
*GM0498	8	MC5A	20U 0128	3	M		PROBAND			Skin biopsy
GM2938A	3	HF12	20U 0185	3	M	WH	PROBAND			Skin biopsy
GM3017	3	HF12	20U 0185	3	M	WH	PROBAND			Skin biopsy

APPARENTLY NORMAL HUMAN FIBROBLAST CELL CULTURES

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
Apparently Normal: Non-Fetal Tissue										
*GM0497	8	EEMEM	20U 0128	4	M	WH	PROBAND			Skin biopsy
*GM0408	8	MC5A	20U 0128	5	F	WH	PROBAND			46,XX; skin biopsy
*GM0409	8	EEMEM	20U 0128	7	M	WH	PROBAND			46,XY; skin biopsy
GM0499	7	MC5A	20U 0128	8	M	WH	PROBAND			Skin biopsy
*GM0038	5	MC5A	20U 0045	9	F	BL	DAU	163	1	46,XX; skin biopsy; mother is GM0043
*GM0500	8	MC5A	20U 0128	10	M	WH	PROBAND			Skin biopsy
GM3348	2	HF12	20U 0026	10	M	WH	PROBAND			Skin biopsy
GM3349	3	HF12	20U 0026	10	M	BL	PROBAND			Skin biopsy
*GM0323	4	MC5A	20U 0125	11	M	WH	BRO	164	1	46,XY; skin biopsy; Sardinian; normal RBC G6PD level, Xg(a) antigen positive; brother of GM0326(XXXXY); parents are GM0321 and GM0322
*GM1582	5	MC5A	20U 0026	11	F	WH	PROBAND	348	1	Formerly GM0240; skin biopsy; tendency to develop translocations in vitro; sister of GM2270
*GM1652	7	MC5A	20U 0133	11	F	WH	PROBAND	165	1	46,XX; skin biopsy; daughter of GM1650 and GM1653, granddaughter of GM1680 and GM1681, sister of GM1651
GM1864	3	HF12	20U 0125	11	M	WH	BRO	161	3	46,XY; normal RBC G6PD level, Xg(a) antigen absent; skin biopsy; brother of GM1863
GM2036	10	EEMEM	10U 0133	11	F	WH	SIS	203	2	Skin biopsy; sister of GM2037
*GM0316A	9	MC5A	20U 0026	12	M	WH	PROBAND			46,XY; skin biopsy
*GM1651	7	EEMEM	20U 0133	13	F	WH	SIS	165	2	46,XX; skin biopsy; daughter of GM1650 and GM1653, granddaughter of GM1680 and GM1681, sister of GM1652
*GM2037	8	EEMEM	10U 0133	13	M	WH	PROBAND	203	1	Skin biopsy; brother of GM2036
*GM0037A	8	MC5A	20U 0045	18	F	WH	MOTHER	162	2	See GM0637 (SV40 transformed GM0037); skin biopsy; Puerto Rican; daughter is GM0041
GM2270	5	HF12	20U 0026	19	M	WH	BRO	348	2	Skin biopsy; brother of GM1582

APPARENTLY NORMAL HUMAN FIBROBLAST CELL CULTURES

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
Apparently Normal: Non-Fetal Tissue										
GM2987	2	HF12	20U 0185	19	M	WH	PROBAND			Skin biopsy
GM3377	2	HF12	20U 0026	19	M	WH	PROBAND			Skin biopsy
GM3440	2	HF12	20U 0185	20	M	WH	PROBAND			Skin biopsy
GM3523	3	HF12	20U 0026	21	M	WH	PROBAND			Skin biopsy
GM2937A	5	HF12	20U 0185	22	M	WH	PROBAND			46,XY; skin biopsy
*GM3234	3	HF12	20U 0215	23	M	AI	PROBAND			Pima Indian; skin biopsy
GM1891	6	HF12	20U 0125	24	M	WH	PROBAND			46,XY; normal RBC level of G6PD(B); skin biopsy
GM3652	3	HF12	20U 0026	24	M	WH	PROBAND			Skin biopsy
GM3651	2	EEMEM	20U 0026	25	F	WH	PROBAND			Skin biopsy
*GM0726	3	MC5A	20U 0026	26	F	OR	PROBAND			Korean; skin biopsy
GM2912A	4	HF12	20U 0185	26	M	WH	PROBAND			Skin biopsy
*GM0495	6	MC5A	20U 0045	29	M		PARENT	417	2	46,XY; formerly GM0104; skin biopsy; clinically normal parent of child with low growth hormone level
GM2674	7	R1640	15U 0030	29	F	WH	PROBAND			Skin biopsy
GM0023	5	MC5A	20U 0095	31	F	WH	PARENT	492	1	46,XX; mother of Down's child; skin biopsy; wife of GM0024
GM0024	5	MC5A	20U 0095	31	M	WH	PARENT	492	2	46,XY; father of Down's child; skin biopsy; husband of GM0023
*GM0043	5	MC5A	20U 0045	32	F	BL	MOTHER	163	2	46,XX; skin biopsy; daughter is GM0038
*GM0964	2	EEMEM	20U 0026	33	M	WH	PROBAND			46,XY; skin biopsy
GM2673	6	MC5A	20U 0030	33	M	WH	PROBAND			Skin biopsy
*GM2185	6	HF12	20U 0186	36	M	WH	PROBAND	455	1	Normal spouse of Huntington's Disease patient; skin biopsy; see GM2184 Lymphoid
GM1650	6	MC5A	20U 0133	37	F	WH	MOTHER	165	3	Skin biopsy; mother of GM1651 and GM1652
GM1653	8	MC5A	20U 0133	37	M	WH	FATHER	165	4	Skin biopsy; father of GM1651 and GM1652, son of GM1680 and GM1681

APPARENTLY NORMAL HUMAN FIBROBLAST CELL CULTURES

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
----Apparently Normal: Non-Fetal Tissue										
GM2665	5	HF12	20U 0147	37	F	WH	PROBAND	524	1	Skin biopsy
GM1717	3	HF12	20U 0125	39	M	WH	PROBAND			46,XY; normal RBC G6PD level, Xg(a) antigen absent; skin biopsy
GM0321	3	MC5A	20U 0125	40	F	WH	MOTHER	164	2	46,XX; skin biopsy; Sardinian; normal RBC G6PD level, Xg(a) antigen positive; sons are GM0323 and GM0326(XXXXY)
GM0322	4	MC5A	20U 0125	40	M	WH	FATHER	164	3	46,XY; skin biopsy; Sardinian; normal RBC G6PD level, Xg(a) antigen positive; sons are GM0323 and GM0326(XXXXY)
*GM0275A	9	EEMEM	20U 0026	42	M	WH	PROBAND			46,XY; skin biopsy
*GM0730	3	MC5A	20U 0026	45	F	WH	PROBAND	343	1	46,XX; skin biopsy; formerly GM0238
GM1863	3	HF12	20U 0125	46	M	WH	PROBAND	161	2	46,XY; normal RBC G6PD level, Xg(a) antigen absent; skin biopsy; brother of GM1864
GM0967	2	EEMEM	20U 0026	51	M	WH	PROBAND	476	1	46,XY; skin biopsy; tendency to develop translocations in vitro
GM3527	5	HF12	20U 0026	55	M	WH	PROBAND			Skin biopsy
GM3526	4	HF12	20U 0026	58	M	WH	PROBAND			Skin biopsy
GM2623	4	HF12	20U 0030	61	F	WH	PARENT			Normal mother of XX male
*GM0288	4	MC5A	20U 0035	64	M	WH	SPOUSE	288	2	Skin biopsy; formerly GM0095; 46,XY; wife is GM0305 (Huntington's disease fibroblast)
GM3529	2	HF12	20U 0026	66	M	BL	PROBAND			Skin biopsy
GM3524	4	HF12	20U 0026	67	F	BL	PROBAND			Skin biopsy
GM3658	4	HF12	20U 0026	68	M	WH	PROBAND	542	1	Skin biopsy; see GM3657 Lymphoid
GM1681	8	MC5A	20U 0133	70	M	WH	PAT GRFR	165	5	Skin biopsy; father of GM1653, grandfather of GM1651 and GM1652
GM1680	8	MC5A	20U 0133	71	F	WH	PAT GRMR	165	6	Skin biopsy; mother of GM1653, grandmother of GM1651 and GM1652
GM3525	3	HF12	20U 0026	80	F	WH	PROBAND			Skin biopsy
*GM1706	6	HF12	20U 0026	82	F	WH	PROBAND	342	1	46,XX; formerly GM0237; skin biopsy

APPARENTLY NORMAL HUMAN FIBROBLAST CELL CULTURES

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
Apparently Normal: Non-Fetal Tissue										
GM3528	3	HF12	20U 0026	83	F	WH	PROBAND			Skin biopsy
GM0731	3	MCSA	20U 0135	96	M	WH	PROBAND			46,XY; skin biopsy

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Alkaptonuria - 20350												
GM2228	4	HF12 20U	0147	36	M	WH	--		PROBAND			Pedigree includes two affected brothers
Argininemia - 20780												
*GM0954	5	EEMEM 20U	0006	4	F	WH	--		PROBAND	422	1	Mexican-American; red cell Arginase deficiency; see GM2011 Lymphoid
Argininosuccinicaciduria - 20790												
*GM0525	8	EEMEM 20U	0123	9	M	WH	--	A	PROBAND	1	1	Argininosuccinase deficiency
*GM0533	8	EEMEM 20U	0123	20	M	WH	--	A	AFF BRO	1	2	Argininosuccinase deficiency
GM0540	8	EEMEM 20U	0123	63	M	WH	+-(0)	B	FATHER	1	3	
GM2830	7	EEMEM 20U	0270	8	F	WH	--		PROBAND	325	1	
GM2831	10	HF12 20U	0270	38	F	WH	+-(0)		MOTHER	325	2	
GM2832	7	MC5A 20U	0270	48	M	WH	+-(0)		FATHER	325	3	
Citrullinuria (Citrullinemia) - 21570												
*GM0063	12	HEMEM 10U	0095	9MO	F	WH	--	A	PROBAND			ATCC CCL 76; deficient Argininosuccinate synthetase activity
GM1684	6	EEMEM 20U	0114	1DA	F	WH	--	A	PROBAND			Neonatal; deficient Argininosuccinate synthetase
GM3056	6	HF12 20U	0134	3	M	WH	--		PROBAND			Mild mental retardation; increased plasma citrulline
*GM1044	3	MC5A 20U	0107	8MO	M	WH	--	A	PROBAND	2	1	See GM1204 Lymphoid; deficient Argininosuccinate synthetase
GM1058	3	MC5A 20U	0107	27	M	WH	+-(0)		FATHER	2	2	See GM1206 Lymphoid
GM1679	2	HF12 20U	0157	1DA	M	WH	--	A	PROBAND	3	1	A sib died of citrullinemia; see also GM1685 Lymphoid; deficient Argininosuccinate synthetase
Cystinosis: Early Onset (Nephropathic Type; Infantile Type) - 21980												
GM0046	5	MC5A 20U	0121	3	M	WH	--	A	PROBAND			Increased intracellular cystine in fibroblasts
GM0706	3	MC5A 20U	0117	18MO	M	WH	--		PROBAND			Pedigree includes an affected sib
GM2066	2	HF12 20U	0019	1	F	WH	--	A	PROBAND			Increased intracellular cystine in fibroblasts
GM0090	3	MC5A 20U	0114	24FW	M	WH	--		PROBAND	4	1	Lung fibroblasts

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Cystinosis: Early Onset (Nephropathic Type; Infantile Type) - 21980												
GM0304	4	MC5A	20U	0114	24FW	M	WH	--		SAME	4	1 Skin fibroblasts; formerly GM0091
GM0489	10	HF12	10U	0114	18MO	M	WH	--	B	AFF BRO	4	2 Fibroblasts show increased intracellular cystine
GM0760	2	MC5A	20U	0128	24FW	M	WH	--		PROBAND	5	1 Fibroblasts also available from bone marrow, kidney, thymus, liver, thyroid, and amnion
GM0018	4	MC5A	20U	0121	3	M	WH	--	A	PROBAND	248	1 Increased intracellular cystine in fibroblasts
GM0020	4	MC5A	20U	0121	5MO	M	WH	--	A	AFF BRO	248	2 Increased intracellular cystine in fibroblasts
*GM0008	5	MC5A	20U	0121	5	F	WH	--	A	PROBAND	478	1 Increased intracellular cystine in fibroblasts
Cystinosis: Benign Type; Adult Type - 21975												
GM0378	8	HF12	10U	0114	7	F	WH	--		PROBAND	6	1 Conjunctiva and cornea contain typical cystinotic crystals
GM0379	8	HF12	10U	0114	4	M	WH	--		AFF BRO	6	2 Conjunctiva and cornea contain typical cystinotic crystals
GM0906	11	HF12	20U	0114	32	M	WH	+-(0)		FATHER	6	3
GM0907	15	HF12	20U	0114	30	F	WH	+-(0)		MOTHER	6	4
Cystinosis: Late Onset Type; Intermediate Type - 21990												
GM0908	8	HF12	20U	0114	12	M	WH	--	B	PROBAND	7	1 Nephropathic cystinosis; fibroblasts show increased levels of intracellular cystine
GM0909	9	HF12	20U	0114	42	F	WH	+-(0)		MOTHER	7	2
GM0910	9	HF12	20U	0114	49	M	WH	+-(0)		FATHER	7	3
Glutaric Aciduria II - 23168												
GM3131		EEMEM	20U	0302	19	F		--		PROBAND		Hypoglycemia and organic aciduria; passage 2 at IMR
Homocystinuria (Cystathionine Synthetase Deficiency) - 23620												
*GM0342	3	MC5A	20U	0066	20	F	WH	--		PROBAND		Excessive urinary homocystine; low IQ; bone pain; seizures; ectopic lens; osteoporosis; spinal compression fractures; B6 responsive

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Homocystinuria (Cystathionine Synthetase Deficiency) - 23620												
GM0423	8	HF12	20U	0114		M		--	A	PROBAND			Def. cystathionine synthetase activity
GM0424A	12	HF12	20U	0114		M		--	A	PROBAND			Def. cystathionine synthetase activity
GM0625	3	MC5A	20U	0038	35	M	WH	+-	A	PARENT			Confirmed heterozygote; two affected children
GM0720	3	MC5A	20U	0038	30	M	WH	--	B	PROBAND			Def. cystathionine synthetase activity
GM0724	14	MC5A	20U	0038		M	WH	--		PROBAND			
GM0864	12	MC5A	20U	0038	13	F		--	A	PROBAND			Def. cystathionine synthetase activity
GM0865	5	MC5A	20U	0038		M		--	A	PROBAND			Def. cystathionine synthetase activity
GM0584	3	MC5A	20U	0038	39	M	WH	--	A	PROBAND	8	1	Def. cystathionine synthetase activity
GM0585	3	MC5A	20U	0038	65	M	WH	+-(0)		FATHER	8	2	
GM0594	2	MC5A	20U	0086	23	M	WH	--	A	PROBAND	9	1	Def. cystathionine synthetase activity
GM0417	4	MC5A	20U	0042	42	F	WH	+-(0)		MOTHER	9	2	
GM0722	7	MC5A	20U	0038	24	M	WH	--	B	PROBAND	10	1	Clinically atypical; formerly GM0548
GM0725	7	MC5A	20U	0038		F	WH	+-(0)		MOTHER	10	2	
GM0751	3	MC5A	20U	0086	19	M	WH	--	A	PROBAND	11	1	Def. cystathionine synthetase activity
GM0752	3	MC5A	20U	0086	23	M	WH	--	A	AFF BRO	11	2	Def. cystathionine synthetase activity
GM0976	3	EEMEM	20U	0086	6	F	WH	+-	A	SIS	11	3	Confirmed heterozygote
GM0753	5	EEMEM	20U	0086	41	M	WH	+-	A	FATHER	11	4	Confirmed heterozygote
GM0813	2	EEMEM	20U	0038	13	M	WH	+-	A	MAT 1ST COU	11	5	Confirmed heterozygote
GM0866	8	MC5A	20U	0038	10	F		--	A	PROBAND	12	1	Def. cystathionine synthetase activity
GM0867	10	EEMEM	20U	0038	6	M		--	A	AFF BRO	12	2	Def. cystathionine synthetase activity
GM0885	3	EEMEM	20U	0086	17	M	WH	--	B	PROBAND	13	1	Def. cystathionine synthetase activity
GM0883	2	EEMEM	20U	0086	42	F	WH	+-(0)		MOTHER	13	2	
GM1128	3	EEMEM	20U	0038	11	F	WH	--	B	PROBAND	14	1	B6 non-responsive
GM1129	3	EEMEM	20U	0038	9	M	WH	--	B	AFF BRO	14	2	B6 non-responsive

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Homocystinuria (Cystathionine Synthetase Deficiency) - 23620												
GM1126	3	EEMEM	20U	0038		M	WH	+-(O)		FATHER	14	3	
GM1374	4	MC5A	20U	0038	13	F	WH	--	A	PROBAND	15	1	Def. cystathionine synthetase activity
GM1375	6	MC5A	20U	0038		F	WH	+-	A	MOTHER	15	2	Confirmed heterozygote
GM1376	6	MC5A	20U	0038		M	WH	+-	A	FATHER	15	3	Confirmed heterozygote
GM0721	7	MC5A	20U	0038	11	F	WH	--	B	PROBAND	639	1	Def. cystathionine synthetase activity; formerly GM0547
Hyperammonemia II: Carbamyl Phosphate Synthetase Deficiency - 23730												
GM3432	8	EEMEM	20U	0049	4MO	F	OT	--		PROBAND			Canadian Rep. No. 615; 46,XX; lethal neonatal hyperammonemia; Pakistani; CPS deficiency confirmed by liver biopsy; positive family history
Hyperglycinemia, Non-Ketotic - 23830												
GM0747	3	MC5A	20U	0107	18MO	F	WH	--		PROBAND			
GM1297	8	HF12	20U	0114		M		--		PROBAND			
GM1301	8	HF12	20U	0114	2MO	F	WH	--		PROBAND			
GM1302	9	HF12	20U	0114	1MO	F		--		PROBAND			
GM0880	5	EEMEM	20U	0095	21	M	WH	--		PROBAND	16	1	Spastic paraparesis
GM1140		MC5A	20U	0095	16	M	WH	--		AFF BRO	16	2	Passage 3 at IMR
GM1162		MC5A	20U	0095	28	M	WH	--		AFF BRO	16	3	Passage 3 at IMR
Hypermethioninemia - 23890												
GM0911	2	EEMEM	20U	0086	3MO	M	WH	--		PROBAND			Deficient hepatic methionine adenosyl-transferase activity; normal activity in fibroblasts
Hyperpipecolatemia - 23940												
*GM3605	7	EEMEM	20U	0134	7	M	WH	--		PROBAND			Hepatomegaly; severe mental retardation; developmental delay; functional blindness; elevated pipecolic acid in serum
Hyperphenylalaninemia - 26160												
GM0006	7	MC5A	20U	0095	5	F	WH	--		PROBAND	17	1	Normal level of dihydropteridine reductase in skin fibroblasts; 46,XX
GM0004	9	MC5A	20U	0095	33	M	WH	+-(O)		FATHER	17	2	46,XY

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Hyperphenylalaninemia - 26160												
GM0007	7	MC5A	20U 0095	30	F	WH	+-(O)		MOTHER	17	3	46, XX; normal level of dihydropteridine reductase in skin fibroblasts
Isovalericacidemia - 24350												
GM0947	5	EEMEM	20U 0002	13	M	WH	--	B	PROBAND			
Maple Syrup Urine Disease - (Branched-Chain Ketoaciduria) - 24860												
*GM0296	15	MC5A	20U 0033	5	M	BL	--	B	PROBAND			Quadriplegic and severely retarded; a brother also has MSUD; 2-5% of normal branched chain amino acid decarboxylase activity
*GM0297	18	MC5A	20U 0033	13DA	F	WH	--	B	PROBAND			Deficient branched chain animo acid decarboxylase activity; high urinary leu., isoleu., and val., and their branched chain alpha-keto acid derivatives
*GM0612	2	MC5A	20U 0074	2MO	F	WH	--	A	PROBAND			Less than 2% of normal branched chain keto-acid decarboxylase activity
GM1744	11	HF12	20U 0077	13DA	F	WH	--	A	PROBAND			Deficient branched chain keto-acid decarboxylase activity
GM1938	5	HF12	20U 0077	5	M	WH	--	A	PROBAND			Deficient branched chain keto-acid decarboxylase activity
*GM0649	3	MC5A	20U 0038	9MO	M	WH	--		PROBAND	18	1	Elevated urine leu., isoleu., and val.; delayed psychomotor development; abnormal EEG
*GM0650	3	MC5A	20U 0038		M	WH	+-(O)		FATHER	18	2	
*GM0651	3	MC5A	20U 0038		F	WH	+-(O)		MOTHER	18	3	
GM1364	2	MC5A	20U 0107	7	F	BL	--	B	PROBAND	20	1	See GM1366 Lymphoid; classical symptoms; one previously affected male sib
GM1654	2	MC5A	20U 0107	5	F	WH	--	A	PROBAND	21	1	See GM1655 Lymphoid; two female sibs died of MSUD; Mennonite kindred; def. branched chain keto-acid decarboxylase
GM1099	3	MC5A	20U 0107	8MO	F	WH	--		PROBAND	266	1	Mennonite; hypotonia; seizures; positive family history
GM2327	3	HF12	20U 0107	4MO	F	WH	--	A	AFF SIS	266	2	Pericardium; deficient branched chain keto-acid decarboxylase; Mennonite

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Maple Syrup Urine Disease - (Branched-Chain Ketoaciduria) - 24860												
GM0016	4	MC5A	20U 0095	25	M	WH	+-	A	PARENT	491	1	Father of an affected child; proband not in Repository; deficient branched chain keto-acid decarboxylase activity; 46,XY
GM0022	5	MC5A	20U 0095	25	F	WH	+-	A	PARENT	491	2	Mother of an affected child; proband not in Repository; deficient branched chain keto-acid decarboxylase activity; 46,XX
Beta-Methylcrotonylglycinuria - 21020 + 21021												
GM3522	2	EEMEM	20U 0128	4MO	M	WH	--		PROBAND			Type II; holocarboxylase synthetase deficiency acidosis; "cat urine" odor; shock
Methylmalonicaciduria - 25100,25110												
GM0050	13	MC5A	20U 0095	1	M	WH	--	A	PROBAND			Unresponsive to B12; ATCC CCL 124; defective methylmalonyl CoA mutase apoenzyme
GM0212	6	MC5A	20U 0095	1	M	WH	--	A	PROBAND			Responsive to B12; cobalamin A mutation
GM0876	5	R1640	20U 0010	1MO	M	WH	--	A	PROBAND			Cobalamin B mutant; B12 responsive
GM0930		HF12	20U 0032	2	F	WH	--	A	PROBAND			Methylmalonyl CoA mutase defect; unresponsive to B12; passage 3 at IMR
GM1673	8	EEMEM	10U 0088	1DA	M	WH	--	B	PROBAND			Apomutase mutant
*GM1674	7	EEMEM	20U 0088	1DA	F	WH	--	B	PROBAND			Cobalamin A mutant; line no. 245
*GM0595	2	MC5A	20U 0088	7	M	WH	--		PROBAND	22	1	Cobalamin A mutant; B12 responsive
GM0596	2	MC5A	20U 0088	25	F	WH	+-(0)		MOTHER	22	2	
GM0306	6	MC5A	20U 0095	2	M		--	A	PROBAND	315	1	Responsive to B12; cobalamin A mutation; formerly GM0051
Methylmalonicaciduria with Homocystinuria - 27740												
GM2452	10	EEMEM	10U 0218	1	M	WH	--	B	PROBAND			cbl D complementation group
GM2453	17	EEMEM	10U 0218	7	F	WH	--	B	PROBAND			cbl C complementation group
Phenylketonuria - 26160												
GM0937	2	EEMEM	20U 0107	19MO	M	WH	--		PROBAND			Normal level of dihydropteridine reductase in skin fibroblasts

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Phenylketonuria - 26160												
GM2406	2	HF12	20U 0074	2MO	M	WH	--		PROBAND			Pyloric stenosis; normal level of dihydropteridine reductase in skin fibroblasts
Propionicacidemia - (Hyperglycinemia with Ketoacidosis & Leukopenia) - 23200												
GM0056	10	MC5A	20U 0088	1	F	WH	--	B	PROBAND			Deficient Propionyl Co A carboxylase activity
GM0057	10	MC5A	20U 0088	6	F	WH	--	B	PROBAND			Deficient Propionyl Co A carboxylase activity
GM1298	8	HF12	20U 0114	4	M		--	B	PROBAND			Deficient Propionyl Co A carboxylase activity
GM1299	7	HF12	20U 0114	2	M		--	B	PROBAND			Deficient Propionyl Co A carboxylase activity
GM1300	8	HF12	20U 0114	30MO	F		--	B	PROBAND			Deficient Propionyl Co A carboxylase activity
GM2805	8	EEMEM	20U 0070	1	M	WH	--		PROBAND			
GM3590	7	EEMEM	20U 0088	22FW	M	WH	--	B	PROBAND			Propionyl Co A carboxylase deficiency; complementation group C
GM0371	3	MC5A	20U 0105	7WK	F	WH	--	B	PROBAND	23	1	Deficient Propionyl Co A carboxylase activity
GM0403	7	MC5A	20U 0105	24	F	WH	+-(0)		MOTHER	23	2	
GM0405	7	MC5A	20U 0105	25	M	WH	+-(0)		FATHER	23	3	
Tyrosinemia - 27670												
GM0286	7	MC5A	20U 0095	2MO	M		--		PROBAND	319	1	Formerly GM0058
DISORDERS OF CARBOHYDRATE METABOLISM												
Amyloidosis - Dominant - 10520												
GM1998	3	HF12	20U 0155	16	F	WH	+-		PROBAND	421	1	Non-Portuguese; see GM1999 Lymphoid
Aspartylglycosaminuria - 20840												
GM0568	2	MC5A	20U 0004	18	F	WH	--		PROBAND			Finnish
*GM2056	3	HF12	20U 0004	32	M	WH	--	B	PROBAND			Finnish; less than 10% of normal N-aspartyl-beta-glycosaminidase activity
*GM2057	4	HF12	20U 0004	27	F	WH	--	B	PROBAND			Finnish; less than 10% of normal N-aspartyl-beta-glycosaminidase activity

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

DISORDERS OF CARBOHYDRATE METABOLISM

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
Aspartylglycosaminuria - 20840												
GM3560	8	EEMEM 20U	0134	10	M	BL	--	B	PROBAND			Deficient N-aspartyl-beta-glycosaminidase; mental retardation and dysmorphic features
Corneal Dystrophy, Macular Type - 21780												
*GM1125	3	EEMEM 20U	0092	41	F	WH	--		PROBAND			Corneal button fibroblast culture; mucopolysaccharidosis type unknown
Fructose-1,6-Diphosphatase Deficiency - 22970												
GM0282	6	MC5A 20U	0095	7	F		--		PROBAND	322	1	Formerly GM0061
Fucosidosis - 23000												
GM0801	6	EEMEM 20U	0138	15	M	WH	--	B	PROBAND			Type II; Spanish-American; alpha-L-fucosidase deficient
GM0802	5	EEMEM 20U	0138	7	M	WH	--	B	PROBAND			Type II; Spanish-American; alpha-L-fucosidase deficient; positive family history
*GM1214	5	EEMEM 20U	0108	18	M	WH	--		PROBAND			Type II; positive family history; alpha-L-fucosidase deficiency in serum and leukocytes
*GM0292	3	MC5A 20U	0078	4	M	WH	--	A	PROBAND	24	1	Type II; deficient alpha-L-fucosidase; 46,XY; see GM1025 Lymphoid
*GM0291	3	MC5A 20U	0078	9	M	WH	--	A	AFF BRO	24	2	See GM1024 Lymphoid; deficient alpha-L-fucosidase
GM0290	3	MC5A 20U	0078	29	F	WH	+-(0)		MOTHER	24	3	See GM1026 Lymphoid
GM0289	3	MC5A 20U	0078	31	M	WH	+-(0)		FATHER	24	4	See GM1023 Lymphoid
Galactosemia (Kinase Deficiency) - 23020												
*GM0334		MC5A 20U	0095	22	F	WH	--	A	PROBAND	25	1	Cataracts; deficient Galactokinase activity; passage 5 at IMR
GM0335		MC5A 20U	0095	53	F	WH	+-(0)		MOTHER	25	2	Passage 3 at IMR
*GM0336		MC5A 20U	0095	62	M	WH	+-(0)		FATHER	25	3	Passage 4 at IMR
Galactosemia (Transferase Deficiency) - 23040												
GM0422		MC5A 20U	0095	9	F	WH	--		PROBAND			Passage 3 at IMR
GM0528		MC5A 20U	0095	2	F	WH	--		PROBAND			Passage 4 at IMR
GM0727	3	MC5A 20U	0095	2MO	F	WH	--		PROBAND			No detectable transferase activity in erythrocytes

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM											
Galactosemia (Transferase Deficiency) - 23040											
*GM1996	6	EEMEM 10U	0002	1	M	WH	+-	A PROBAND			Duarte variant-galactosemia heterozygote; reduced transferase activity in fibroblasts and erythrocytes
*GM0052	11	EEMEM 10U	0095	8	M	BL	--	B PROBAND	26	1	G6PD type B; ATCC CCL 133; see also GM0639 (SV40 transformed) and GM2796 Lymphoid; no detectable transferase activity in RBCs or fibroblasts
GM1908	2	MC5A 20U	0095	15	M	BL	--	B SAME	26	1	Earlier passage number
*GM0053	12	EEMEM 10U	0095	6	M	BL	--	B AFF BRO	26	2	G6PD Type A; ATCC CCL 132; no detectable transferase activity in RBCs or fibroblasts
*GM0054	10	MC5A 20U	0095	3MO	M	WH	--	B PROBAND	27	1	See also GM0638 (SV40 Transformed) and GM2795 Lymphoid; G6PD type B
GM1907	3	MC5A 20U	0095	6	M	WH	--	B SAME	27	1	Earlier passage number
*GM0441	6	MC5A 20U	0069	15	M	WH	--	A PROBAND	28	1	See GM2412 Lymphoid; no detectable transferase activity
*GM0440	6	MC5A 20U	0069	16	F	WH	--	A AFF SIS	28	2	See GM1028 Lymphoid; no detectable transferase activity
*GM0439	6	MC5A 20U	0069	41	F	WH	+-(0)	B MOTHER	28	3	See GM1029 Lymphoid; 50% of normal transferase activity in erythrocytes
*GM0438	6	MC5A 20U	0069	46	M	WH	+-(0)	B FATHER	28	4	50% of normal transferase activity in erythrocytes
*GM0442	6	MC5A 20U	0069	12	M	WH	++	B BRO	28	5	Normal transferase activity in erythrocytes
*GM1533	7	MC5A 20U	0069	19	F	WH	++	B SIS	28	6	Normal transferase and kinase activity in fibroblasts
GM1209	2	EEMEM 20U	0045	9MO	M	WH	--	A PROBAND	29	1	Identical (single placenta); no detectable transferase activity
GM1210	2	EEMEM 20U	0045	9MO	M	WH	--	A AFF TWIN	29	2	Identical (single placenta); no detectable transferase activity
GM1211	3	EEMEM 20U	0045	4	M	WH	+-	A BRO	29	3	50% of normal transferase activity
GM1212	3	EEMEM 20U	0045	29	F	WH	+-	A MOTHER	29	4	50% of normal transferase activity

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Galactosemia (Transferase Deficiency) - 23040												
GM1208	3	EEMEM 20U	0045	27	M	WH	+-(O)		FATHER	29	5	
GM1417	6	MC5A 20U	0095	22	F		--		PROBAND	30	1	Slow growing culture; no detectable transferase activity in erythrocytes
GM1418	5	MC5A 20U	0095		F		+-(O)		MOTHER	30	2	Slow growing culture; carrier level of transferase in erythrocytes
GM1419	5	MC5A 20U	0095		M		+-(O)		FATHER	30	3	Slow growing culture; carrier level of transferase in erythrocytes
GM1703	4	HF12 20U	0095	9MO	M	WH	--	B	PROBAND	31	1	2nd generation with GALT
GM1704	3	HF12 20U	0095	2	M	WH	--	B	AFF COU	31	2	2nd generation with GALT
GM1741	8	HF12 20U	0077	24FW	M	WH	--	B	PROBAND	32	1	See GM1743 Amniotic
*GM0433	3	MC5A 20U	0095	10	F	BL	--	A	PROBAND	300	1	Fibroblasts accumulate large amounts of intracellular gal-1-phosphate and galactitol
Glycogen Storage Disease: Type I - Von Gierke Disease - 23220												
GM0574	8	EEMEM 20U	0130	25	M	WH	--		PROBAND			Absent hepatic glucose-6-phosphatase
GM3719	9	EEMEM 10U	0006	5	M	OT	--		PROBAND			Type Ib; Panamanian Ind.;severe hypoglycemia, lactic acidosis, & neutropenia; liver bx shows G6P trans. defect
Glycogen Storage Disease: Type II - Pompe Disease - 23230												
GM0244		MC5A 20U	0002	5MO	F	WH	--	A	PROBAND			Mexican; deficient acid-alpha-1,4 glucosidase; passage 4 at IMR; 46,XX
*GM0248		MC5A 20U	0095	5MO	M	BL	--	A	PROBAND			Deficient acid-alpha-1,4 glucosidase; passage 3 at IMR
GM0338	2	MC5A 20U	0008	1MO	M	BL	--	B	PROBAND			Culture grows poorly; absent acid-alpha-1,4 glucosidase
*GM0443		MC5A 20U	0099	30	M		--	A	PROBAND			Late onset; 23% of normal acid-alpha-1,4 glucosidase activity; passage 3 at IMR
GM1935	3	MC5A 20U	0126	30	F	BL	--	A	PROBAND			Adult onset; deficient acid-alpha-1,4 glucosidase
GM3329	2	HF12 20U	0107	23MO	M	BL	--	A	PROBAND			Deficient acid-alpha-1,4 glucosidase
Glycogen Storage Disease: Type III - Debranching Enzyme Deficiency - 23240												
GM0111	8	MC5A 20U	0143	1	F	WH	--	A	PROBAND			46,XX; less than 10% of normal debrancher enzyme activity

HUMAN FIBROBLAST CULTURES WITH
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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Glycogen Storage Disease: Type III - Debranching Enzyme Deficiency - 23240												
GM0226	12	MC5A	20U 0143	13	F		--	A	PROBAND			Less than 10% of normal debrancher enzyme activity
GM0573	7	EEMEM	20U 0062	16	F	WH	--	B	PROBAND			Deficient debrancher enzyme activity
GM0576	6	EEMEM	20U 0062	7	F	OR	--	B	PROBAND			Chinese; deficient debrancher enzyme activity
GM0578	18	EEMEM	20U 0062	4	M	WH	--	B	PROBAND			Latin-American; deficient debrancher enzyme activity
GM1702	4	HF12	20U 0095	11	M		--		PROBAND			
GM3388	7	HF12	20U 0077	18MO	M	WH	--	A	PROBAND			Arab; decreased amylo-1-6 glucosidase activity in fibroblast
GM3390	6	HF12	20U 0077	16MO	M	WH	--	A	PROBAND			Arab; deficient amylo-1-6-glucosidase activity
GM0303	9	MC5A	20U 0095	9	F	WH	--	A	PROBAND	317	1	Formerly GM0055; less than 10% of normal debrancher enzyme activity
Glycogen Storage Disease: Type IV - Andersen Disease - 23250												
GM0572	9	EEMEM	20U 0062	2	F	WH	--	A	PROBAND			Brancher enzyme deficiency
Glycogen Storage Disease: Type V - McArdle Disease - 23260												
GM0577	7	EEMEM	20U 0130	39	M	WH	--		PROBAND			Absent muscle phosphorylase activity; normal activity in fibroblasts
Glycogen Storage Disease: Type VI - Hers Disease - 23270												
GM0579	9	EEMEM	20U 0062		M		--		PROBAND			Low hepatic phosphorylase activity; normal activity in fibroblasts
Glycogen Storage Disease: Type VIII, IX - P'ase Kinase Deficiency - 30600												
GM0025	4	MC5A	20U 0077	30MO	M	WH	y-	A	PROBAND			46,XY; deficient D-phosphophosphorylase kinase
GM0575	5	EEMEM	20U 0062	3	M	WH	y-	B	PROBAND			Mexican-American; less than 10% of normal phosphorylase b-kinase activity; type IX
Glycogen Storage Disease: Type Unclassified												
GM2523	3	HF12	20U 0107	18MO	M	BL	--		PROBAND			Debrancher enzyme deficiency in muscle & low phosphorylase activity in liver
Hyperpyruvicacidemia												
GM0583	3	MC5A	20U 0038	3MO	F	WH						

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Leigh Encephalomyelopathy - 25600												
GM1503	2	MC5A 20U	0045	42MO	F	WH	--		PROBAND			Infantile subacute
GM3672	2	EEMEM 20U	0105	16MO	F	WH	--		PROBAND			Developmental regression; microcephalic; elevated blood lactic acid & pyruvate; only affected family member; 46,XX
Mannosidosis - 24850												
*GM0654	3	MC5A 20U	0004	7	M	WH	--	A	PROBAND			Deficient alpha-mannosidase; Finnish
*GM1851	10	MC5A 20U	0006	3	M	WH	--	B	PROBAND			Deficient alpha-mannosidase
GM2817	14	EEMEM 15U	0062	6	F	WH	--	A	PROBAND			22% of normal mannosidase activity
GM2051	18	HF12 20U	0077	7	F	WH	--	B	PROBAND	33	1	Variant; Arab; approximately 20% of normal alpha-mannosidase activity
GM2050	9	HF12 20U	0077	11	M	WH	--	B	AFF BRO	33	2	Variant; Arab; approximately 20% of normal alpha-mannosidase activity
GM2049	8	HF12 10U	0077	39	M	WH	+-(0)	B	FATHER	33	3	Arab; approximately 50% of normal alpha-mannosidase activity
Mucolipidosis: Type I (Lipomucopolysaccharidosis) - 25240												
*GM2685	11	EEMEM 20U	0230	14	M	WH	--	A	PROBAND			Zero to 12% normal neuraminidase activity; normal B-glucuronidase and B-hexosaminidase levels
Mucolipidosis: Type II - I-Cell Disease - 25250												
GM1742	3	HF12 20U	0077	4MO	F	WH	--		PROBAND			Arab; elevated alpha-mannosidase & beta-hexosaminidase in serum; culture grows poorly
GM3112	7	HF12 20U	0095	21FW	F	WH	--		PROBAND			
*GM0080	5	MC5A 20U	0095	16	F	WH	+-(0)		MOTHER	34	2	46,XX; two affected children
*GM0081	5	MC5A 20U	0095	23	M	WH	+-(0)		FATHER	34	3	46,XY; two affected children
GM3066	5	HF12 20U	0260	23FW	F	WH	--		PROBAND	34	4	
GM1589	7	HF12 20U	0077		M	WH	+-(0)		PARENT	35	2	Father of an affected child
GM1590	7	HF12 20U	0077		F	WH	+-(0)		PARENT	35	3	Mother of an affected child
GM2013	8	HF12 30U	0077	1MO	M	WH	--	B	PROBAND	36	1	Arab; fibroblasts show deficiencies for several lysosomal hydrolases
GM2014	8	HF12 20U	0077	18	F	WH	+-(0)		MOTHER	36	2	Arab

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Mucolipidosis: Type II - I-Cell Disease - 25250												
GM2045	11	MC5A	10U 0095	1DA	M	WH	--	B	PROBAND	37	1	Fibroblasts show deficiencies for several lysosomal hydrolases
GM2046	6	HF12	20U 0095	25	F	WH	+-(0)		MOTHER	37	2	
GM2047	4	MC5A	20U 0095	26	M	WH	+-(0)		FATHER	37	3	
GM2273	3	HF12	20U 0070	2	M	WH	--		PROBAND	38	1	Low levels of activity for several lysosomal enzymes
GM2274	3	HF12	20U 0070		F	WH	+-(0)		MOTHER	38	2	
Mucolipidosis: Type III - Pseudo-Hurler Polydystrophy - 25260												
*GM0113	4	MC5A	20U 0105	30MO	F	WH	--	A	PROBAND			50% of normal levels of Alpha-L-Iduronidase, beta-glucuronidase, and beta-galactosidase
*GM1494	6	MC5A	20U 0057	16	F	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase, beta-glucuronidase, and beta-galactosidase
*GM1759	2	HF12	20U 0092	13	M	WH	--	A	PROBAND			Similarly affected sister; low levels of beta-galactosidase, beta-glucuronidase, beta-hexosaminidase, alpha-fucosidase, & alpha-mannosidase
*GM2065	3	HF12	20U 0126	9	M	WH	--	A	PROBAND			Deficient beta-galactosidase, hexosaminidase, arylsulfatase A, and beta-glucuronidase
*GM2425	6	EEMEM	20U 0134	15	M	WH	--	B	PROBAND			Elevated serum acid hydrolases; excessive inclusion bodies in cytoplasm; deficient fibroblast neuraminidase activity
*GM3685	4	HF12	20U 0126	15	M	WH	--	B	PROBAND			Abnormal mucopolysaccharide accumulation and deficiencies for multiple acid hydrolases in fibroblasts
GM2559	2	HF12	20U 0126	7	F	BL	--		PROBAND	501	1	
GM2558	2	HF12	20U 0126	9	F	BL	--	B	AFF SIS	501	2	40% of normal beta-galactosidase & 15% of normal beta-hexosaminidase activity
GM3391	8	HF12	20U 0077	15	M	WH	--	B	PROBAND	610	1	Iranian; deficient in Alpha-L-Iduronidase, arylsulfatase A, alpha-mannosidase and hexosaminidase A & B
GM3392	7	HF12	20U 0077	13	F	WH	--	B	AFF SIS	610	2	Iranian; deficient in Alpha-L-Iduronidase, arylsulfatase A, alpha-mannosidase and hexosaminidase A & B

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Mucolipidosis: Type IV - 25265												
GM2525	22	HF12	20U 0077	2	F	WH	--	B	PROBAND			Ashkenazi
GM2526	15	HF12	20U 0077	2	F	WH	--	B	PROBAND			Ashkenazi
GM2527	4	HF12	20U 0077	20MO	M	WH	--	B	PROBAND			Ashkenazi
*GM2528	20	HF12	20U 0077	8MO	M	WH	--	B	PROBAND			Ashkenazi; fibroblasts show grossly abnormal storage bodies & normal levels of acid hydrolases
GM2629	9	HF12	20U 0077	2	M	WH	--		PROBAND			
*GM2048	10	HF12	20U 0077	2	F	WH	--	B	PROBAND	229	1	Ashkenazi; see GM2533 Lymphoid; affected sib is GM2529 Amniotic; photophobia; bilateral corneal opacities; abnormal lysosomal storage bodies
Mucopolysaccharidosis: Type IH - Hurler Syndrome - 25280												
*GM0415	2	MC5A	20U 0107	4	M	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase
GM0887	5	EEMEM	20U 0057	7	F	WH	--	A	PROBAND			No detectable Alpha-L-Iduronidase activity
GM1053	5	EEMEM	20U 0057	4	M	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase; culture grows slowly
GM1257	7	EEMEM	15U 0062	1	F	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase; Latin-American
*GM0002	7	MC5A	20U 0095	10MO	M	WH	--	A	PROBAND	39	1	Deficient Alpha-L-Iduronidase; accumulates excess mucopolysaccharides; correctable by Hurler factor; 46,XY
GM0003	7	MC5A	20U 0095	26	F	WH	+-(0)		MOTHER	39	2	46,XX
*GM0034	6	MC5A	20U 0095	9MO	F	WH	--	A	PROBAND	40	1	Deficient Alpha-L-Iduronidase; 46,XX
GM0031	4	MC5A	20U 0095	23	F	WH	+-(0)		MOTHER	40	2	46,XX
GM0042	4	MC5A	20U 0095	25	M	WH	+-(0)		FATHER	40	3	46,XY
GM0798		EEMEM	20U 0062	1	F	WH	--	A	PROBAND	41	1	Deficient Alpha-L-Iduronidase; passage 3 at IMR
GM0799		EEMEM	20U 0062	22	F	WH	+-(0)		MOTHER	41	2	Passage 3 at IMR
GM0800		EEMEM	20U 0062	27	M	WH	+-(0)		FATHER	41	3	Passage 3 at IMR

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DISORDERS OF CARBOHYDRATE METABOLISM

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
Mucopolysaccharidosis: Type IH - Hurler Syndrome - 25280												
*GM1391	2	MC5A 20U	0070	9MO	F	WH	--	A	PROBAND	42	1	Deficient Alpha-L-Iduronidase
GM1392	3	MC5A 20U	0070	24	F	WH	+-(0)		MOTHER	42	2	
GM1393	2	MC5A 20U	0070	24	M	WH	+-(0)		FATHER	42	3	
Mucopolysaccharidosis: Type IH/S - Hurler/Scheie - 25280												
GM0512	4	MC5A 20U	0008	15	F	IN	--	A	PROBAND			Deficient Alpha-L-Iduronidase
*GM0963	3	EEMEM 20U	0092	5	M	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase
GM2846		MC5A 20U	0096	14	F	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase; similarly affected sister; passage 5 at IMR
GM1254	9	EEMEM 20U	0062	4	M	WH	--	A	PROBAND	43	1	Deficient Alpha-L-Iduronidase
GM1255	6	EEMEM 15U	0062	10	M	WH	--	A	AFF BRO	43	2	Deficient Alpha-L-Iduronidase
GM1898	10	HF12 20U	0077	6	M	WH	--	B	PROBAND	44	1	Arab; deficient Alpha-L-Iduronidase
GM2016	6	HF12 10U	0077	35	F	WH	+-(0)		MOTHER	44	2	Arab
GM2017	6	HF12 20U	0077	40	M	WH	+-(0)		FATHER	44	3	Arab
GM2845	3	MC5A 20U	0096	23	M	WH	--	A	PROBAND	326	1	Deficient Alpha-L-Iduronidase
GM2847	3	MC5A 20U	0096	25	F	WH	--	A	AFF SIS	326	2	Deficient Alpha-L-Iduronidase
Mucopolysaccharidosis: Type IS - Scheie Syndrome - 25310												
GM1256	9	EEMEM 15U	0062	12	M	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase
GM1323	3	MC5A 20U	0092	58	M	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase
Mucopolysaccharidosis: Type II - Hunter Syndrome - 30990												
GM0039	4	MC5A 20U	0066	9	M	WH	y-	A	PROBAND			Accumulates mucopolysaccharide, correctable by Hunter factor; deficient iduronate sulfatase activity; 46,XY
GM0047	6	MC5A 20U	0107	9	M	WH	y-	A	PROBAND			Accumulates mucopolysaccharide, correctable by Hunter factor
GM0140	4	EEMEM 20U	0105	5	M	BL	y-	A	PROBAND			Excessive mucopolysaccharide accumulation correctable by Hunter factor
GM0298	4	MC5A 20U	0066	2	M	WH	y-	A	PROBAND			Similarly affected brother; excessive mucopolysaccharide accumulation correctable by Hunter factor

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Mucopolysaccharidosis: Type II - Hunter Syndrome - 30990												
*GM0690	3	MC5A	20U	0092	14	M	WH	y-	A	PROBAND			Deficient iduronate sulfatase activity
GM0862	2	EEMEM	20U	0107	11	M	WH	y-	A	PROBAND			Mild type; similarly affected half brother; Puerto Rican; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM1258	7	EEMEM	15U	0062	2	M	WH	y-	A	PROBAND			Excessive mucopolysaccharide accumulation correctable by Hunter factor
GM1929	5	EEMEM	20U	0096	7	M	WH	y-	B	PROBAND			Severe type; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM2268	8	MC5A	20U	0168	5	F	WH	-?	A	PROBAND			Rare Hunter's in female; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM0614	3	MC5A	20U	0092	18	M	WH	y-	A	PROBAND	45	1	Mild type; deficient iduronate sulfatase activity
*GM0613	3	MC5A	20U	0092		F	WH	+-(0)	A	MOTHER	45	2	Normal mucopolysaccharide accumulation; positive family history for Hunters
*GM0615	3	MC5A	20U	0092	9	M	WH	y-	A	PROBAND	46	1	Severe type; deficient iduronate sulfatase activity
*GM0620	3	MC5A	20U	0092		F	WH	+-(0)	A	MOTHER	46	2	Normal mucopolysaccharide accumulation; has had two affected male children
GM1928	5	EEMEM	20U	0096	7	M	WH	y-	B	PROBAND	48	1	Mild type; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM1927	5	EEMEM	20U	0096	16	M	WH	y-	B	AFF UNCLE	48	2	Mild type; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM1583	7	HF12	20U	0077	3	M	WH	y-	A	PROBAND	247	1	Israeli; affected brother is GM1584 Amniotic; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM1896	9	HF12	20U	0088	20FW	M	WH	y-	B	PROBAND	597	1	Same patient as GM1794; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM3181A	3	HF12	20U	0170	22	M	WH	y-	A	PROBAND	601	1	Excess mucopolysaccharide accumulation, correctable by Hunter factor; two similarly affected cousins

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DISORDERS OF CARBOHYDRATE METABOLISM

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
Mucopolysaccharidosis: Type II - Hunter Syndrome - 30990											
GM3180A	3	HF12	20U	0170	27	F	WH +?	A SIS	601	2	Normal mucopolysaccharide accumulation; possible heterozygote
Mucopolysaccharidosis: Type IIIA - Sanfilippo Syndrome, A - 25290											
GM0312	2	MC5A	20U	0107	3	F	WH --	A PROBAND			Deficient heparan sulfate sulfatase
GM0629	3	MC5A	20U	0066	10	M	--	A PROBAND			Deficient heparan sulfate sulfatase, normal alpha-D-Glucosaminidase
GM0643	3	MC5A	20U	0107	3	M	WH --	A PROBAND			Deficient heparan sulfate sulfatase
GM0934	2	EEMEM	20U	0070	7	F	WH --	A PROBAND			Deficient heparan sulfate sulfatase
GM0879	7	EEMEM	20U	0057	3	F	WH --	A PROBAND	49	1	Deficient heparan sulfate sulfatase
GM0886	7	EEMEM	20U	0057	19	F	WH +-(0)	MOTHER	49	2	
GM1094	3	MC5A	20U	0095	5	F	WH --	A PROBAND	50	1	Deficient heparan sulfate sulfatase, normal N-acetyl-alpha glucosaminidase
GM1739	3	MC5A	20U	0095	6	F	WH --	A SAME	50	1	
GM1095	4	MC5A	20U	0095		F	WH +-(0)	MOTHER	50	2	
GM1096	3	MC5A	20U	0095		M	WH +-(0)	FATHER	50	3	
Mucopolysaccharidosis: Type IIIB - Sanfilippo Syndrome, B - 25292											
GM0156	5	MC5A	20U	0107	7	M	WH --	A PROBAND			No detectable N-acetyl-alpha-D glucosaminidase activity; 46,XY
GM0737	3	MC5A	20U	0107	7	M	WH --	A PROBAND			Deficient N-acetyl-alpha-glucosaminidase
GM2552	3	HF12	20U	0105	7	F	WH --	A PROBAND			Deficient N-acetyl-alpha-glucosaminidase; increased urinary heparan sulfate
GM2931	5	HF12	20U	0077	3	F	WH --	PROBAND			Ashkenazi
GM1426	3	MC5A	20U	0070	1	F	WH --	A PROBAND	289	1	Deficient N-acetyl-alpha-D glucosaminidase; similarly affected brother
Mucopolysaccharidosis: Type IV - Morquio Syndrome - 25300											
*GM0593	3	MC5A	20U	0092	7	F	WH --	A PROBAND			Less than 1% of normal N-acetylgalactosamine 6-sulfate sulfatase, normal B-galactosidase, B-glucuronidase activity
GM0958	2	EEMEM	20U	0092	12	M	WH --	A PROBAND			Clinically atypical; 3% of normal N-acetylgalactosamine 6-sulfate sulfatase activity

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF CARBOHYDRATE METABOLISM								
Mucopolysaccharidosis: Type IV - Morquio Syndrome - 25300												
GM1259	9	EEMEM 15U	0062	14	F	WH	--	A	PROBAND			Clinically atypical; culture grows poorly; deficient N-acetylgalactosamine 6-sulfate sulfatase, beta-galactosidase, and beta-glucuronidase
GM1361	3	MC5A 20U	0092	43	M	WH	--	A	PROBAND			Variant; 3% of normal N-acetylgalactosamine 6-sulfate sulfatase activity, normal B-galactosidase & B-glucuronidase
Mucopolysaccharidosis: Type IVB-Beta-Galactosidase Deficient Morquio - 25301												
GM3251	6	EEMEM 20U	0134	4	F	WH	--	B	PROBAND			7% of normal B-galactosidase activity, normal N-acetylgalactosamine 6-sulfate sulfatase activity
Mucopolysaccharidosis: Type IV - Morquio Syndrome - type unknown												
GM1602	3	MC5A 20U	0066	11	F	WH	--	A	PROBAND	222	1	Normal level of N-acetylgalactosamine 6-sulfate sulfatase; normal beta-galactosidase (low normal)
GM2455	3	HF12 20U	0066	6	F	WH	--	A	AFF SIS	222	2	Normal level of N-acetylgalatosamine 6-sulfate sulfatase; normal beta-galactosidase (low normal)
GM2456	3	HF12 20U	0066	30	M	WH	+-(O)		FATHER	222	3	
GM2555	4	HF12 20U	0066	30	F	WH	+-(O)		MOTHER	222	4	
Mucopolysaccharidosis: Type VI - Maroteaux-Lamy Syndrome - 25320												
GM2849	3	HF12 20U	0070	35	M	WH	--	B	PROBAND			Deficient Arylsulfatase B
*GM0519	9	MC5A 20U	0008	4	F	WH	--	A	PROBAND	51	1	See GM1022 Lymphoid; deficient Arylsulfatase B
*GM0520	8	MC5A 20U	0008	42	M	WH	+-	A	FATHER	51	2	Heterozygote level of Arylsulfatase B
*GM0935	6	EEMEM 20U	0008	40	F	WH	+-	A	MOTHER	51	3	Heterozygote level of Arylsulfatase B
*GM0943	7	EEMEM 20U	0008	8	F	WH	+-	A	SIS	51	4	Heterozygote level of Arylsulfatase B
*GM0942	7	EEMEM 20U	0008	5	F	WH	++	A	SIS	51	5	Normal enzyme level
GM0538	9	MC5A 20U	0105	18MO	F	WH	--	B	PROBAND	267	1	Deficient Arylsulfatase B, normal Alpha-L-Iduronidase
GM2572	3	HF12 20U	0105	2	M	WH	--	B	AFF BRO	267	2	
GM3720	6	EEMEM 20U	0016	25	F	BL	+-(O)		MOTHER	392	1	See GM3721 Lymphoid

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DISORDERS OF CARBOHYDRATE METABOLISM												
Mucopolysaccharidosis: Type VI - Maroteaux-Lamy Syndrome - 25320												
GM3722	3	HF12	20U 0016	40MO	F	BL	--		PROBAND	392	2	5% of normal Arylsulfatase B activity; see GM3723 Lymphoid
GM3724	5	HF12	20U 0016	41	M	BL	+-(O)		FATHER	392	3	See GM3725 Lymphoid
GM3726	3	HF12	20U 0016	9	F	BL	+?		SIS	392	4	Possible heterozygote; see GM3727 Lymphoid
Mucopolysaccharidosis: Type VII - Beta-Glucuronidase Deficiency - 25322												
GM3303	5	MC5A	20U 0126	14DA	M		--	B	PROBAND			Deficient beta-glucuronidase
*GM0121	4	MC5A	20U 0126	3	M	BL	--	A	PROBAND	52	1	2% of normal B-glucuronidase, normal A-galactosidase, acid B-galactosidase, and A-mannosidase; 46,XY
GM1850	3	HF12	20U 0126	28	M	BL	+-	A	FATHER	52	2	Carrier enzyme level
GM2074	4	HF12	20U 0126	21	F	BL	+-(O)		MOTHER	52	3	
*GM0151	3	MC5A	20U 0066	4	M	WH	--	A	PROBAND	425	1	Deficient beta-glucuronidase
GM2784	4	HF12	20U 0066	3MO	F	WH	--	B	AFF SIS	425	2	No detectable beta-glucuronidase activity in fibroblasts
Neuraminidase (Sialidase) Deficiency - 25655												
*GM0806	3	MC5A	20U 0126	30MO	F	WH	--	A	PROBAND			Deficient beta-galactosidase and sialidase activity
GM2837	6	MC5A	20U 0134	31	M	WH	--	A	PROBAND			22% of normal sialidase activity
GM3589	8	EEMEM	20U 0126	7MO	F	WH	--	B	PROBAND			Less than 1% of normal beta-galactosidase activity; deficient sialidase activity; both parents have 50% of normal beta-galactosidase activity
*GM1718	2	HF12	20U 0070	2MO	F		--	A	PROBAND	53	1	Less than 1% of normal neuraminidase activity
GM1719	3	HF12	20U 0070	25	F		+-(O)		MOTHER	53	2	
GM1720	2	HF12	20U 0070	26	M		+-(O)		FATHER	53	3	
GM2921	9	EEMEM	20U 0134	2	M	WH	--	A	PROBAND	330	1	6 times the normal level of sialic acid in fibroblasts; approx. 8% of normal sialidase activity
GM2922	11	EEMEM	20U 0134		M	WH	+-	B	FATHER	330	2	58% of normal sialidase activity using NAN-lactose as substrate

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DISORDERS OF CARBOHYDRATE METABOLISM												
Neuraminidase (Sialidase) Deficiency - 25655												
GM2923	9	EEMEM 20U	0134	29	F	WH	+-	B	MOTHER	330	3	62% of normal sialidase activity using NAN-lactose as substrate
Phosphoglycerate Kinase Deficiency - 31180												
*GM0743	2	MC5A 20U	0098	4	M	OR	y-		PROBAND			Japanese; nonspherocytic hemolytic anemia; cerebral palsy; mental deficiency; decreased muscle & erythrocyte PGK activity; variant PGK (Matsue)
Tricarboxylic Acid Cycle, Defect of - 27537												
GM3093	9	EEMEM 20U	0088	4	F	WH	--	B	PROBAND			Pyruvate dehydrogenase complex deficiency
DISORDERS OF LIPID METABOLISM												
Beta-sitosterolemia - 21025												
GM1995	6	MC5A 20U	0072	9	M	WH	--		PROBAND			Xanthomas
GM1994	12	MC5A 20U	0072	11	M	WH	--		PROBAND	352	1	Xanthomas; moderate hypercholesterolemia
GM1600	5	MC5A 20U	0072	18	F	WH	--		AFF SIS	352	2	Tendon xanthomas; slightly elevated cholesterol
GM1599	6	MC5A 20U	0072	42	M	WH	+-(O)		FATHER	352	3	Diabetic
GM1601	5	MC5A 20U	0072	41	F	WH	+-(O)		MOTHER	352	4	Clinically normal
Myopathy: Carnitine-palmityl-transferase Deficiency - 25513												
GM1763		MC5A 20U	0095	29	M	WH	--		PROBAND	344	1	Formerly GM0249; recurrent myoglobinuria; passage 4 at IMR
Cholesterol Ester Storage Disease of Liver - 21500												
GM3111	6	R1640 20U	0187	22	M	WH	--		PROBAND			Liver lysosomal acid lipase <5% of control and electrophoresis showed absence of "A" band
GM3347	8	EEMEM 20U	0006	7	F	WH	--	B	PROBAND			Mexican; approximately 5% of control level of acid lipase activity
*GM0863	5	EEMEM 20U	0006	11	F	WH	--	A	PROBAND	381	1	Latin American; two similarly affected sisters; less than 5% of normal acid lipase activity in fibroblasts
Fabry Disease (Diffuse Angiokeratoma) - 30150												
GM0107	5	MC5A 20U	0105	10	M		y-	A	PROBAND			27% of normal level of alpha-galactosidase; normal B-glucur., acid-B-galact., and A-mannosidase

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM											
Fabry Disease (Diffuse Angiokeratoma) - 30150											
*GM0881	9	EEMEM 20U	0108	17	M	WH	y-	B PROBAND			About 20% of normal alpha-galactosidase activity; conjunctiva showed lipid deposits in the endothelial cells of the vessel walls
*GM0882	10	EEMEM 20U	0108	40	M	WH	y-	B PROBAND			Italian; history of proteinuria and mild fatigability; about 30% of normal alpha-galactosidase activity
*GM2769	7	MC5A 20U	0238	18	M	WH	y-	A PROBAND	246	1	Alpha gal A absent, Alpha gal B normal, Xg(a) antigen negative
*GM2774	3	MC5A 20U	0238	47	F	WH	+-	A AUNT	246	3	50% of normal Alpha gal A, normal Alpha gal B, Xg(a) antigen +/-
*GM2775	3	MC5A 20U	0238	16	M	WH	y-	A AFF MATICOU	246	4	Alpha gal A absent, alpha gal B normal, Xg(a) antigen positive
*GM2771	6	HF12 20U	0238	42	F	WH	+-	A AUNT	246	5	50% of normal Alpha gal A, normal Alpha gal B, Xg(a) antigen +/-
*GM2772	4	MC5A 20U	0238	38	F	WH	+-(0)	A AUNT	246	7	50% of normal Alpha gal A, normal Alpha gal B, Xg(a) antigen +/-
Farber Lipogranulomatosis - 22800											
GM2315	8	MC5A 20U	0175	7	F	WH	--	B PROBAND			Italian descent; a sister with similar clinical history died at age 18; deficient acid ceramidase
GM2314	4	MC5A 20U	0175	6	F	WH	--	B PROBAND	56	1	Irish descent; deficient acid ceramidase
GM2316	3	MC5A 20U	0175	37	M	WH	+-(0)	FATHER	56	2	Irish descent
GM2317	3	MC5A 20U	0175	37	F	WH	+-(0)	MOTHER	56	3	Irish descent
GM1 Gangliosidosis: Type I - 23050											
*GM0918	18	EEMEM 20U	0049	1	M		--	A PROBAND			From Canadian Repository; zero beta-galactosidase activity; 46,XY
GM1 Gangliosidosis: Type III - 23065											
GM2438	12	EEMEM 20U	0049	9MO	F		--	B PROBAND			From Canadian Repository; 20% of normal B-galactosidase activity
GM2439	16	EEMEM 20U	0049				--	B PROBAND			From Canadian Repository; deficient B-galactosidase activity
GM2 Gangliosidosis: Tay-Sachs Disease, AB Variant - 27275											
GM1675	5	EEMEM 20U	0008	16MO	F	BL	--	PROBAND			

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM												
GM2 Gangliosidosis: Type I - Tay-Sachs Disease - 27277, 27280												
*GM0077	9	MC5A	20U	0095	1	M	BL	--	A	PROBAND			Non-Jewish; deficient hexosaminidase A
*GM0221	4	MC5A	20U	0066	3	M	WH	--	A	PROBAND			Deficient hexosaminidase A
*GM0502	2	MC5A	20U	0066	11MO	M	WH	--	A	PROBAND			Jewish; deficient hexosaminidase A
GM0527	3	MC5A	20U	0107	15MO	M	WH	--	A	PROBAND			Deficient hexosaminidase A
GM0515	5	EEMEM	20U	0068	1	F	WH	--	A	PROBAND	58	1	Pericardium fibroblast culture; deficient hexosaminidase A; Jewish
GM0514	8	EEMEM	20U	0068	3	F	WH	--	B	AFF SIS	58	2	Pericardium fibroblast culture; Jewish; deficient hexosaminidase A
GM1110	7	EEMEM	20U	0068	10MO	M	WH	--	A	PROBAND	59	1	Non-Jewish; electrophoresis shows absence of hexosaminidase A band
GM1109	7	EEMEM	20U	0068	31	M	WH	+-(0)		FATHER	59	2	Non-Jewish
GM1108	8	MC5A	20U	0068	30	F	WH	+-(0)		MOTHER	59	3	Non-Jewish
GM2968		HF12	20U	0008	12MO	F	WH	--	A	PROBAND	437	1	Ashkenazi; no detectable hexosaminidase A activity by electrophoresis; passage 4 at IMR
GM3051	8	HF12	20U	0008	23	M	WH	+-	B	FATHER	437	2	Ashkenazi
GM3052	7	HF12	20U	0008	24	F	WH	+-	B	MOTHER	437	3	Ashkenazi; same patient as GM2969
GM2 Gangliosidosis: Type II - Sandhoff Disease - 26880												
*GM0294	5	R1640	20U	0017	2	M	WH	--	A	PROBAND			Deficient hexosaminidase A & B
*GM0317	3	MC5A	20U	0066	1	F	BL	--	A	PROBAND			Deficient hexosaminidase A & B
*GM0470	8	EEMEM	20U	0009	1	F	WH	--	A	PROBAND			Deficient hexosaminidase A & B
*GM0203	5	MC5A	20U	0107	1	M	WH	--	A	PROBAND	60	1	Deficient hexosaminidase A & B; 46,XY
GM0204	5	MC5A	20U	0107	18	F	WH	+-(0)		MOTHER	60	2	46,XX
GM2094	2	HF12	20U	0181	12	M	WH	--	B	PROBAND	432	1	Juvenile; cerebellar ataxia; severe mental retardation; deficient hexosaminadase A and B; same patient as GM2144
Gaucher Disease: Type 2 (Infantile, Cerebral) - 23090												
GM1260	2	EEMEM	20U	0050	11MO	F	WH	--	A	PROBAND			<10% of normal fibroblast glucosylceramide Beta-glucosidase activity; splenomegaly; opisthotonos; died age 1 yr

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM												
Gaucher Disease: Type 2 (Infantile, Cerebral) - 23090												
GM2627	2	HF12	20U 0107	3	F	WH	--		PROBAND			Atypical; subacute neuropathic Gauchers; myoclonus; seizures; died age 3 due to pneumonia
*GM0877	2	EEMEM	20U 0107	1	M	WH	--	A	PROBAND	57	1	< 5% normal fibroblast glucosylceramide B-glucosidase activity; strabismus, trismus; hepatosplenomegaly; died age 1
GM0878	2	EEMEM	20U 0107		F	WH	+-	A	MOTHER	57	2	Carrier level of fibroblast glucosyl-ceramide B-glucosidase activity
Gaucher Disease: Type 3 (Juvenile and Adult, Cerebral) - 23100												
GM0372	4	MC5A	20U 0009	29	M	WH	--		PROBAND			Adult
*GM0852	5	EEMEM	20U 0138	20	M	WH	--	B	PROBAND			Adult; beta-glucosidase deficiency
GM1607	3	MC5A	20U 0047	30	M	WH	--	A	PROBAND			Adult; eastern European Jewish; deficient B-glucosidase; hepato-splenomegaly since age 17
Familial Hypercholesterolemia - 14389 & Hyperlipoproteinemia: Type II-14440												
*GM1915	4	HF12	20U 0050	13	F	BL	--	A	PROBAND			Receptor negative
GM0283	5	MC5A	20U 0045	28	F	WH	+-		MOTHER	61	1	Formerly GM0102; elevated plasma cholesterol
GM0376	5	MC5A	20U 0045	6	F	WH	+-		PROBAND	61	2	Formerly GM0099; elevated plasma cholesterol
GM0488	4	MC5A	20U 0084	11	M	WH	--	A	PROBAND	62	1	Receptor negative; approx. 10 fold increase in HMG CoA reductase in fibroblasts
GM0483	4	MC5A	20U 0084	37	F	WH	+-		MOTHER	62	2	Elevated plasma cholesterol
GM2000	2	HF12	20U 0050	7	F	WH	--	A	PROBAND	63	1	Receptor negative; same patient as GM1116
*GM0701	7	HF12	20U 0072	6	F	WH	--	A	PROBAND	64	1	Receptor negative; approx. 10 fold increase in HMG CoA reductase in fibroblasts
GM0700	7	HF12	20U 0072	45	F	WH	+-		MOTHER	64	2	
*GM2408A	2	HF12	20U 0050	14	M		--	A	PROBAND	272	1	Normal receptor binding with defective receptor internalization; line J.D.
GM1355	3	MC5A	20U 0093	13	F	WH	-?	A	PROBAND	274	1	25 to 40% of normal LDL receptor activity; possibly a severe heterozygote, mild homozygote, or double heterozygote

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DISORDERS OF LIPID METABOLISM												
Familial Hypercholesterolemia - 14389 & Hyperlipoproteinemia: Type II-14440												
GM1354	3	MC5A	20U 0093	38	F	WH	+-		MOTHER	274	2	Elevated plasma cholesterol; no xanthomas
GM1385	3	MC5A	20U 0093	38	M	WH	+-		FATHER	274	3	Elevated plasma cholesterol; no xanthomas
GM1386	2	MC5A	20U 0093	17	M	WH	++		BRO	274	4	Normal plasma cholesterol
*GM0486	4	MC5A	20U 0084	25	F	WH	--	A	PROBAND	387	1	Receptor negative; see GM1459 Lymphoid
GM3040	11	EEMEM	20U 0072	9	M	WH	--	B	PROBAND	440	1	Receptor negative or defective; very high serum LDL; normal triglycerides; cutaneous and tendinous xanthomas; corneal arcus
GM3064	6	EEMEM	20U 0072	29	M	WH	+-		FATHER	440	2	No xanthomas; no clinical evidence of atherosclerosis; elevated plasma cholesterol
GM3065	6	EEMEM	20U 0072	30	F	WH	+-		MOTHER	440	3	No xanthomas; no clinical evidence of atherosclerosis; family history is strongly positive for early atherosclerosis and hypercholesterolemia
Familial Hypercholesterolemia - Variants												
*GM0667	5	HF12	20U 0072	12	M	WH	--		PROBAND	263	1	Lebanese; elevated plasma cholesterol; pedigree shows autosomal recessive inheritance
*GM0694	6	HF12	20U 0072	15	F	WH	--		AFF SIS	263	2	Lebanese; elevated plasma cholesterol; xanthomas
GM0695	6	HF12	20U 0072	55	M	WH	+-(O)		FATHER	263	3	Lebanese
*GM0696	6	HF12	20U 0072	5MO	F	WH	--		AFF SIS	263	4	Lebanese; elevated plasma cholesterol; xanthomas
Krabbe Disease (Globoid Cell Sclerosis) - 24520												
GM1773	2	HF12	20U 0105	1	M	WH	--	A	PROBAND			Deficient lactosylceramide B-galactosidase
GM0267	7	R1640	20U 0058	36	F	WH	+-	A	PARENT	97	1	Proband not in Repository; 35% of normal galactocerebroside beta-galactosidase activity
GM0268	6	R1640	20U 0058	41	M	WH	+-	A	PARENT	97	2	Proband not in Repository; 45% of normal galactocerebroside beta-galactosidase activity

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DISORDERS OF LIPID METABOLISM												
Krabbe Disease (Globoid Cell Sclerosis) - 24520												
GM3045	2	HF12	20U 0008	19FW	F		--		PROBAND	442	1	
GM3046A		HF12	20U 0008	19FW	F		--		IDENT TWIN	442	2	Affected; one placenta, one sac; passage 4 at IMR
Metachromatic Leukodystrophy: Adult - 25000												
GM0132	7	MC5A	20U 0105	22	M	WH	+-	B	1ST COU	66	1	Heterozygote levels of arylsulfatase A
GM2093	3	HF12	20U 0105	25	M	WH	--	B	PROBAND	66	3	Deficient arylsulfatase A
Metachromatic Leukodystrophy: Infantile - 25010												
*GM0078	7	MC5A	20U 0131	3	M	WH	--	A	PROBAND			Deficient arylsulfatase A
GM0243	2	MC5A	20U 0074	3	M	WH	--	A	PROBAND			Deficient arylsulfatase A
GM0905	2	EEMEM	20U 0122	3	F	WH	--	A	PROBAND			Deficient arylsulfatase A
GM2331	5	MC5A	20U 0134	5	M	WH	--	A	PROBAND			Deficient arylsulfatase A
*GM0197	3	MC5A	20U 0105	4	M	WH	--	A	PROBAND	65	1	Deficient arylsulfatase A
GM0196	3	MC5A	20U 0105	33	M	WH	+-(0)		FATHER	65	2	46,XY; three children affected with infantile MLD
*GM0200	4	MC5A	20U 0105	5	M	WH	+-	B	BRO	65	4	46,XY; carrier level of arylsulfatase A
GM0357		MC5A	20U 0105	3	M	WH	+-	B	MAT 1ST COU	65	6	Carrier level of arylsulfatase A; passage 4 at IMR
GM0640	10	MC5A	20U 0105	25	F	WH	+-	B	MAT AUNT	65	8	Mother of GM0357; carrier level of arylsulfatase A
Multiple Sulfatase Deficiency Disease - 27220												
*GM2407	8	MC5A	20U 0214	3	F	WH	--	A	PROBAND	237	1	Deficient arylsulfatase A,B,CC, sulfamidase, iduronate-sulfate sulfatase, GalNAc-6 and GlcNAc-6-sulfate sulfatase
GM2644	7	HF12	20U 0214	22	F	WH	+-(0)		MOTHER	237	2	
GM2667	7	EEMEM	20U 0214	27	M	WH	+-(0)		FATHER	237	3	
*GM3245	3	HF12	20U 0105	9	M	WH	--	B	PROBAND	632	1	See GM3246 Lymphoid; deficient arylsulfatase A, B, and C
Niemann-Pick Disease: Type A - 25720												
GM0112	3	MC5A	20U 0143	10MO	M	WH	--	A	PROBAND			No detectable sphingomyelinase activity

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF LIPID METABOLISM								
Niemann-Pick Disease: Type A - 25720												
GM0370	8	EEMEM 15U	0002	13MO	F	WH	--	A	PROBAND			7% of normal sphingomyelinase activity
*GM0406	9	MC5A 20U	0105	21MO	F	WH	--	A	PROBAND			8% of normal sphingomyelinase activity
GM0559		MC5A 20U	0105	25	F	WH	+-	B	PROBAND			Jewish; about 20% of normal sphingomyelinase activity; positive family history; carrier; passage 4 at IMR
GM0641	19	MC5A 20U	0105	4	M	WH	--	B	PROBAND			Deficient sphingomyelinase activity
Niemann-Pick Disease: Type B - 25720												
GM3252	3	MC5A 20U	0095	21FW	F	WH	--	A	PROBAND			2% of normal sphingomyelinase activity; similarly affected brother
GM3393	8	EEMEM 20U	0006	10	F	WH	--	B	PROBAND			Non-Jewish; less than 5% of normal sphingomyelinase activity
Niemann-Pick Disease: Type C - 25720												
*GM0110A	6	MC5A 20U	0107	9	M	WH	--	A	PROBAND			Variant; 24% of normal sphingomyelinase activity
GM3123	2	HF12 20U	0107	9	F	WH	--	B	PROBAND	451	1	See GM3124 Lymphoid; 38% of normal sphingomyelinase activity
Niemann-Pick Disease: Type Unspecified												
GM0165	3	MC5A 20U	0107	7	M	WH	--	A	PROBAND			Clinically atypical; 27% of normal sphingomyelinase activity
GM2895	6	HF12 20U	0073	2	M	WH	--	A	PROBAND	328	1	47% of normal sphingomyelinase activity; juvenile or variant form; Puerto Rican
GM2896	5	MC5A 20U	0073	33	F	WH	+-	B	MOTHER	328	2	50% of normal sphingomyelinase activity
GM2897	8	MC5A 20U	0073	25	M	WH	+-	B	FATHER	328	3	50% of normal sphingomyelinase activity
Niemann-Pick Disease without Sphingomyelinase Deficiency - 25725												
GM3519	14	EEMEM 20U	0333	29	M	WH	--	A	PROBAND			Normal sphingomyelinase activity; not Jewish or Nova Scotian; two similarly affected male sibs; sea blue histiocytes in bone marrow
Schilder Disease - 26910 + 30010												
GM0269	4	MC5A 20U	0058	30	F	WH	+?		PARENT			Possible carrier for fatal X-linked Schilder's disease with adrenal insufficiency; one affected son
*GM2448	7	EEMEM 15U	0175	8	M	WH	--		PROBAND			Adrenoleukodystrophy; mild neurological impairment - not clearly progressive; negative family history; four normal maternal uncles

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM												
Sea-Blue Histiocyte Disease - 26960												
GM0843	3	EEMEM 20U	0066	10	F	WH	--		PROBAND	121	1	
GM0844	3	EEMEM 20U	0066		M	WH	--		AFF BRO	121	2	
GM1912	3	HF12 20U	0348	24	F	WH	--		PROBAND	407	1	See GM1913 Lymphoid; muscle biopsy showed foamy histiocytes & sea-blue histiocytes; eye movement abnormalities
Wolman Disease - 27800												
*GM1606	4	HF12 20U	0154	7MO	F		--	A	PROBAND			Deficient lysosomal acid lipase activity
GM2121	7	MC5A 20U	0187	31	F	WH	+-	B	PARENT	462	1	Fibroblasts have 50% of normal lysosomal acid lipase activity; proband (GM2211) is a poor grower and is not listed in catalog
*GM2109	11	MC5A 20U	0178	3MO	M	BL	--		PROBAND	631	1	Fatty metamorphosis of liver with cirrhosis & cholestasis; foamy histiocytic infil.of spleen, lymph nodes, lamina propria of sm. intestine & bone marrow
GM3557	6	EEMEM 20U	0178	24	F	BL	+?	B	MOTHER	631	2	Possible carrier levels of acid lipase; 46,XX
GM3558	5	EEMEM 20U	0178	28	M	BL	+?	B	FATHER	631	3	Normal level of acid lipase; 46,XY
DISORDERS OF METAL METABOLISM												
Menkes Syndrome (Kinky Hair Disease) - 30940												
*GM0220	4	MC5A 20U	0107	2MO	M	WH	y-	A	PROBAND	79	1	Fibroblasts exhibit elevated Cu levels
*GM1057	3	MC5A 20U	0107	5DA	M	WH	y-	A	AFF BRO	79	2	See GM1245 Lymphoid; fibroblasts exhibit elevated copper concentrations; 46,XY
*GM0245	3	MC5A 20U	0074	1MO	M	WH	y-	A	AFF COU	79	3	Fibroblasts exhibit elevated Cu levels; 46,XY
*GM1981	3	HF12 20U	0107	2	M	WH	y-	A	PROBAND	80	1	See GM1982 Lymphoid; fibroblasts exhibit elevated copper concentrations
GM1983	3	HF12 20U	0107	26	F	WH	+?		MOTHER	80	2	See GM1984 Lymphoid; son is only affected child in an extended family
GM3700	2	HF12 20U	0078	9MO	M	WH	y-		PROBAND	390	1	Low serum copper and ceruloplasmin
GM3659A	5	EEMEM 20U	0078	24	F	WH	+?		MOTHER	390	2	Two previous first trimester spontaneous abortions; son is only affected individual in a large pedigree

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DISORDERS OF METAL METABOLISM												
Wilson Disease (Hepatolenticular Degeneration) - 27790												
*GM0032	4	MC5A	20U	0095	9	F	WH	--	PROBAND			Asymptomatic, patient on penicillamine; fibroblasts show elevated intracellular copper concentration
*GM0033	4	MC5A	20U	0128	16	M	WH	--	PROBAND			Fibroblasts show elevated intracellular copper concentration
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Adenine Phosphoribosyltransferase Deficiency - 10260												
*GM0517		EEMEM	20U	0071	64	M	BL	+-	PROBAND			Enzyme deficient in erythrocytes; normal level in fibroblasts; passage 3 at IMR
Adenosine Deaminase Deficiency, with Immunodeficiency - 24275												
*GM0469	8	MC5A	20U	0118	1	M	WH	--	B	PROBAND		About 1% of normal ADA activity
*GM0471	4	HF12	20U	0103	18MO	F	BL	--		PROBAND		Two male sibs are similarly affected; no detectable RBC ADA
GM1390	5	HF12	20U	0090	3MO	M	WH	--	A	PROBAND		Short-limbed dwarfism
*GM2444	14	MC5A	20U	0208	8	F	WH	--	A	PROBAND		French-Canadian; deficient ADA activity in RBC and fibroblasts; similarly affected male sib
GM2446	6	MC5A	20U	0208		F	WH	--	A	PROBAND		Deceased; Spanish-American
GM3136		MC5A	20U	0237	8MO	F	WH	--	A	PROBAND		Passage 2 at IMR; Sephardic Jew; classical early onset severe form; RBC's, fibroblasts, & mononuclear cells show ADA deficiency
GM3137		MC5A	20U	0237		F		--	A	PROBAND		Passage 2 at IMR; classical early onset severe form; deficient ADA activity in erythrocytes, lymphocytes, & fibroblasts
*GM2027	9	MC5A	20U	0118		F		+-	B	PARENT	202	2 Mother of ADA deficient child; clinically normal; 53% of normal ADA activity in fibroblasts
*GM2028	11	MC5A	20U	0118		M		+-	B	PARENT	202	3 Father of ADA deficient child; clinically normal; 48% of normal ADA activity in fibroblasts
*GM2434	6	HF12	20U	0208	7MO	M	WH	--	A	PROBAND	219	1 Deficient RBC and fibroblast ADA activity; similarly affected female sib

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DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Adenosine Deaminase Deficiency, with Immunodeficiency - 24275												
*GM2433	6	HF12	20U 0208	25	F	WH	+-	A	MOTHER	219	2	Clinically normal; carrier levels of ADA activity in RBC, lymphocytes, and fibroblasts
GM2605	2	HF12	20U 0223	8MO	M	WH	--	A	PROBAND	226	1	See GM2606 Lymphoid
GM2607	3	HF12	20U 0223		F	WH	+-	A	MOTHER	226	2	See GM2608 Lymphoid
GM2609	2	HF12	20U 0223		M	WH	+-	A	FATHER	226	3	See GM2610 Lymphoid
GM2578	2	HF12	20U 0223	5MO	M	WH	--	A	PROBAND	232	1	RBC, lymphocytes ADA approx 1% of normal levels
GM2580	4	HF12	20U 0223	38	M	WH	+-	A	FATHER	232	2	See GM2581 Lymphoid; carrier level of ADA activity in fibroblasts
GM2582	3	HF12	20U 0223	37	F	WH	+-	A	MOTHER	232	3	See GM2583 Lymphoid; carrier level of ADA activity in fibroblasts
GM2436	5	MC5A	20U 0208	NB	F	WH	--	A	PROBAND	388	1	Swiss; deceased; deficient ADA activity in fibroblasts
GM2435	5	HF12	20U 0208	25	F	WH	+-	A	MOTHER	388	2	Swiss; clinically normal; carrier level of fibroblast ADA activity
GM2447	11	MC5A	20U 0208	25	M	WH	+-	A	FATHER	388	3	Swiss; clinically normal; carrier level of fibroblast ADA activity
GM2445	8	MC5A	20U 0217	3	M	BL	--	A	PROBAND	423	1	See GM1715 Lymphoid; deficient ADA activity in fibroblasts
GM2824	3	HF12	20U 0291	4MO	F	WH	--		PROBAND	446	1	See GM2825A Lymphoid; deficient ADA activity in RBC and lymphocytes
GM1997	7	MC5A	20U 0208	4MO	M	BL	--		PROBAND	633	1	See GM2294 Lymphoid; deficient ADA activity in RBC
Adenosine Deaminase Deficiency, with no Immunodeficiency - 24276												
*GM3029	5	HF12	20U 0170	16	M	BL	--	B	PROBAND	634	1	Kung South African; see GM3043 Lymphoid; clinically normal
Inosine Triphosphate Pyrophosphohydrolase (ITPase) Deficiency - 14752												
GM1617	2	HF12	20U 0177	29	F	WH	+-		PROBAND	635	1	See GM1619 Lymphoid; deficient RBC ITPase
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800												
GM0068	14	EEMEM	10U 0027	2	M		y-	A	PROBAND			Fibroblasts nonviable in HAT medium

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DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM											
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800											
GM0152	10	HF12	20U 0101		M	WH	y-	A PROBAND			
GM0537	7	HF12	20U 0031	13	M		y-	A PROBAND			
*GM1362	3	MC5A	20U 0066	10	M	WH	y-	A PROBAND			Fibroblasts nonviable in HAT medium
*GM2330	9	EEMEM	10U 0039	20	M	WH	y-	B PROBAND			Variant; normal intelligence; no self-mutilation; HPRT activity in fibroblasts is reduced but not absent; cells survive in HAT medium
*GM3467	10	HF12	20U 0331	5DA	M	WH	y-	B PROBAND			Fibroblasts resistant to azaguanine & thioguanine; cannot utilize exogenous hypoxanthine; HPRT deficient
GM0377	8	MC5A	20U 0095	8MO	M	WH	y-	A PROBAND	69	1	No detectable HPRT activity; formerly GM0029
GM0013	7	MC5A	20U 0095	34	F	WH	+-	A MOTHER	69	2	46,XX; 55% of normal HPRT activity in fibroblasts
*GM0158	9	HF12	20U 0085	3	M	BL	y-	A PROBAND	71	1	G6PD type A
*GM0135	8	EEMEM	20U 0101		F	BL	+-	A MOTHER	71	2	Sister of GM0318, HPRT+, G6PDB/HPRT-, G6PDA; 46,XX
*GM0318	13	EEMEM	20U 0101	40	F	BL	+-	A MAT AUNT	71	3	HPRT+, G6PDB/HPRT-,G6PDA; formerly GM0134
*GM0159	11	HF12	20U 0085	10	M	BL	y-	A AFF MAT COUS	71	4	G6PD type A
*GM2063	10	EEMEM	10U 0101	5	M	BL	y-	A AFF MAT COUS	71	6	G6PD Type A; see also GM0847 (SV40 Transformed);formerly GM0177
*GM1662	2	HF12	20U 0066	9	M	WH	y-	A PROBAND	72	1	No detectable HPRT, normal APRT activity
GM1659	3	HF12	20U 0066	42	F	WH	+-	MOTHER	72	2	Normal blood HPRT; carrier by hair root assay
GM1661	3	HF12	20U 0066	14	F	WH	+-	SIS	72	3	Normal blood HPRT; carrier by hair root assay
GM1660	3	HF12	20U 0066	11	F	WH	+-	SIS	72	4	Normal blood HPRT; carrier by hair root assay
*GM2227	3	HF12	20U 0066	12	M		y-	A PROBAND	73	1	No detectable HPRT; normal APRT activity
*GM2226	3	HF12	20U 0066	40	F		+?	MOTHER	73	2	

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800												
*GM2290	2	HF12	20I	0183	19FW	M	WH	y-	A	PROBAND	74	1	Fetal skin fibroblast culture; see also GM2292 Lymphoid and GM2338 Amniotic
*GM2291	2	HF12	20U	0183	19FW	M	WH	y-	A	SAME	74	1	Fetal lung fibroblast culture; see also GM2292 Lymphoid and GM2338 Amniotic
*GM3116	2	HF12	20U	0183	19FW	M	WH	y-	B	AFF BRO	74	2	Fetal kidney fibroblast culture
*GM3117	1	HF12	20U	0183	19FW	M	WH	y-	B	AFF BRO	74	2	Fetal lung fibroblast culture; same fetus as GM3116
GM2851	3	HF12	20U	0066	15	M		y-	A	PROBAND	508	1	Same mother as GM2852; deficient HPRT; normal APRT activity
GM2852	2	HF12	20U	0066	7	M		y-	A	HALF BRO	508	2	Deficient HPRT; normal APRT activity
Myo-adenylate Deaminase Deficiency - 25475												
GM3071	4	EEMEM	20U	0178	14	M	WH	--		PROBAND	444	1	Skin fibroblast culture; muscle aching in all 4 extremities; elevated serum CPK; similarly affected 1st cousin; no detectable muscle ADA activity
GM3073	5	EEMEM	20U	0178	38	F	WH	+-(O)		MOTHER	444	2	Skin fibroblast culture; 40% of normal muscle ADA activity
GM3075	5	EEMEM	20U	0178	58	M	WH	+-(O)		FATHER	444	3	Skin fibroblast culture; 70% of normal muscle ADA activity
Orotic Aciduria - 25890												
*GM0328		EEMEM	20U	0080	2	M	WH	--	B	PROBAND			Culture medium should contain uridine; line AUC; passage 2 at IMR
GM0632	2	EEMEM	20U	0115	14	F	WH	--	B	PROBAND			Culture medium should contain uridine
GM1406	2	MC5A	20U	0257		F	WH	--		PROBAND			Medium should contain uridine; orotic acid crystals in urine; megaloblastic anemia with normal B12 & folic acid levels; pt. responded to oral uridine
Xeroderma Pigmentosum: Dominant Inheritance - 19440												
GM2881	8	EEMEM	20U	0294	67	M	WH	+-		PROBAND			First skin tumor at age 34; 4 of 4 siblings affected as well as two nephews
Xeroderma Pigmentosum: Complementation Group A - 27870												
*GM0544	2	MC5A	20U	0023	10	M	WH	--		PROBAND			De Sanctis-Cacchione; XP4LO
*GM0710	11	MC5A	20U	0011	18MO	M	WH	--	B	PROBAND			De Sanctis-Cacchione; XP25RO; ATCC CRL 1261

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DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Complementation Group A - 27870												
*GM2994	3	HF12	20U	0023	4	M	WH	--	A	PROBAND			Egyptian; CNS symptoms; XP5CA
*GM2990	2	HF12	20U	0023	8	F	WH	--	A	PROBAND	468	1	Egyptian; XP1CA
*GM2991	3	HF12	20U	0023	6	M	WH	--	A	AFF 1ST COU	468	2	Egyptian; XP2CA
Xeroderma Pigmentosum: Complementation Group B - 27870												
GM1854	3	HF12	20U	0155	64	F	WH	+-(0)		MOTHER	283	2	Mother of GM2252 Lymphoid; see also GM1855 Lymphoid; also heterozygous for Cockayne syndrome
Xeroderma Pigmentosum: Complementation Group C - 27870												
*GM0709	17	MC5A	20U	0011	15	M	WH	--	B	PROBAND			XP21RO; Dutch
*GM2992	3	HF12	20U	0023	13	F	WH	--	A	PROBAND			Egyptian; XP3CA
*GM2997	3	HF12	20U	0023	4	M	WH	--	A	PROBAND			Egyptian; consanguinous parents; XP7CA
GM0671	4	MC5A	20U	0023	15	M	WH	--		PROBAND	75	1	XP8BE; ATCC CRL 1158; see GM2249 Lymphoid
GM0676	3	MC5A	20U	0023	15	M	WH	--		AFF TWIN	75	2	XP9BE; ATCC CRL 1161; see GM2498 Lymphoid
GM0673	4	MC5A	20U	0023	21	M	WH	--		AFF BRO	75	3	XP4SL
*GM0677	3	MC5A	20U	0023	25	M	WH	--		AFF BRO	75	4	XP2BE; ATCC CRL 1166
*GM2993	3	HF12	20U	0023	8	F	WH	--	A	PROBAND	466	1	Egyptian; XP4CA; consanguinous parents
*GM2998	2	HF12	20U	0023		F	WH	+-(0)		MOTHER	466	2	Egyptian; XPH4CA
*GM2995	3	HF12	20U	0023	10	M	WH	--	A	PROBAND	467	1	Egyptian; XP6CA
*GM2996	2	HF12	20U	0023	7WK	M	WH	--	A	AFF BRO	467	2	Egyptian; XP8CA; no clinical symptoms of XP at time of biopsy but fibroblasts show defective DNA repair
*GM0522	6	EEMEM	20U	0037	16	F	BL	--		PROBAND	503	1	G6PD type B; Phosphoglucomutase 1; XP2NBi
GM0523A	9	MC5A	20U	0037	10	F	BL	--	A	AFF SIS	503	2	G6PD type B; Phosphoglucomutase 1; XP1NBi
GM0030	3	MC5A	20U	0110	24	M	WH	--	B	PROBAND	507	1	XP3BE; ATCC CRL 1189; see GM2248 Lymphoid
*GM2096	3	HF12	20U	0007	15	F	BL	--		PROBAND	627	1	De Sanctis-Cacchione; Lupus erythematosus; see GM2634 Lymphoid; XP1MI; 46,XX

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DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Complementation Group D - 27870												
GM0436	3	MC5A	20U 0023	22	F	WH	--		PROBAND			XP1NE; near normal unscheduled DNA synthesis
GM3615	11	MC5A	20U 0023		F		--		PROBAND			XP1BR
*GM0434	3	MC5A	20U 0023	28	M	WH	--		PROBAND	502	1	De Sanctis-Cacchione; XP3NE; 30 to 50% of normal unscheduled DNA synthesis
*GM0435	3	MC5A	20U 0023	23	F	WH	--		AFF SIS	502	2	De Sanctis-Cacchione; XP2NE; 30 to 50% of normal unscheduled DNA synthesis
Xeroderma Pigmentosum: Complementation Group E - 27870												
*GM2415	12	HF12	20U 0011	34	F	WH	--	B	PROBAND	626	1	XP2RO; ATCC CRL 1259; relative of XP3RO (GM2450 Lymphoid)
Xeroderma Pigmentosum: Complementation Group F - 27870												
GM3542	9	HF12	20U 0335	29	M	OR	--	B	PROBAND			Japanese; XP3YO; 10% of normal post UV unscheduled DNA synthesis; no neurological abnormalities
Xeroderma Pigmentosum: Complementation Group G - 27870												
*GM3021A	13	HF12	20U 0011	17	F		--	B	PROBAND			XP2BI
GM3616	10	MC5A	20U 0023		M		--		PROBAND			XP3BR
Xeroderma Pigmentosum: Complementation Group Undetermined - 27870												
GM0510	9	EEMEM	10U 0023	10	M	WH	--	A	PROBAND			XP1PW; Arab; low unscheduled DNA synthesis
*GM0936	8	EEMEM	20U 0046	22FW	F	WH	--		PROBAND			Umbilical cord fibroblast culture; low in unscheduled DNA synthesis
GM1213	2	EEMEM	20U 0133	61	F	WH	--		PROBAND			Diagnosed at age 34
GM1227	3	MC5A	20U 0133	28	M	WH	--	A	PROBAND			Diagnosed at age 13; low repair activity
GM1295	4	EEMEM	20U 0145	6	F	WH	--		PROBAND			XP1KC; extreme photosensitivity, photophobia, and rhinorrhea; negative family history
GM1509	3	MC5A	20U 0133	38	F	WH	--		PROBAND			
GM2004	3	HF12	20U 0023	30	F	WH	--		PROBAND			Patient shows gradual mental retardation; XP1WA; 46,XX; 50% of cells have unidentified 2q+
GM2033	4	HF12	20U 0170	16	M	BL	--		PROBAND			XP3JO; Zulu

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DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Complementation Group Undetermined - 27870												
GM2060	8	MC5A	20U	0133	33	F	WH	--	A	PROBAND			Deficient post UV unscheduled DNA synthesis
GM2062	6	MC5A	20U	0133	3	M	WH	--		PROBAND			XP5PD; 46,XY
GM2476	2	HF12	20U	0170	10	F	BL	--		PROBAND			Zulu; XP6JO
GM3083	6	EEMEM	20U	0133	54	F	WH	+-(O)		PARENT			Mother of an affected child
*GM3617	11	MC5A	20U	0023		M		--	B	PROBAND			XP30RO; variant; light to moderate XP symptoms; 88% of control level of unscheduled DNA synthesis
GM3618	7	MC5A	20U	0023		F		--		PROBAND			XP6DU; variant
GM0082	5	MC5A	20U	0044	12	F	WH	--		PROBAND	76	1	Classical X.P. symptoms
GM0241	11	MC5A	20U	0044		F	WH	+-(O)		MOTHER	76	2	
GM1630	2	MC5A	20U	0173	42MO	M	WH	--	A	PROBAND	77	1	Deficient repair; XP1WI; see GM2344 Lymphoid
GM1631	2	MC5A	20U	0173	29	M	WH	+-	A	FATHER	77	2	Near normal repair
GM1632	3	EEMEM	20U	0173	25	F	WH	+-	A	MOTHER	77	3	Near normal repair
*GM2032	2	HF12	20U	0170	5	M	BL	--	B	PROBAND	78	1	XP2JO; Zulu; tongue tumor fibroblast culture
*GM2035	3	HF12	20U	0170	5	M	BL	--	B	SAME	78	1	XP2JO; Zulu; skin fibroblast culture
*GM2034	4	HF12	20U	0170	28	F	BL	+-(O)		MOTHER	78	2	XPH2JO; Zulu
*GM2024	2	HF12	20U	0170	3	M	BL	--		AFF BRO	78	3	XP1JO; Zulu
GM2009	5	EEMEM	10U	0056	10	F	WH	--		PROBAND	200	1	XP1PD; mental and growth retardation; photosensitivity
GM2010	7	EEMEM	10U	0056	4	M	WH	--		AFF BRO	200	2	XP2PD; mental and growth retardation; photosensitivity
GM1389	4	MC5A	20U	0133	21	F	WH	--		PROBAND	377	1	See GM1646 Lymphoid; diagnosed at age 16 yrs
GM3081A	6	EEMEM	20U	0133	51	F	WH	+-(O)		PARENT	415	3	Mother of an affected child
*GM2359	3	HF12	20U	0046	25	M	WH	--		PROBAND	433	1	Variant; DNA excision repair normal; DNA post-replication repair is abnormal; Iranian; XP115LO; same patient as GM2111; parents are first cousins

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DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Complementation Group Undetermined - 27870												
*GM3055	6	EEMEM 20U	0023	28	M	WH	--	B	PROBAND	438	1	XP1CH; variant; 60% of normal unsched. DNA synthesis; parents are first cousins
*GM3054	9	EEMEM 20U	0023	50	F	WH	+-(O)	B	MOTHER	438	3	XPH1CH; Russian Armenian; normal unscheduled DNA synthesis and repair replication
GM3614	3	HF12 20U	0110	21	F	BL	--		PROBAND	495	1	Microcephaly, retarded growth, mental deficiency; sickle cell trait; hypo- and hyperpigmented skin areas
GM3176	3	HF12 20U	0023	3	F	WH	--		PROBAND	600	1	XP1AA
GM3177	2	HF12 20U	0023	2	M	WH	+?		BRO	600	2	Clinically unaffected; possible heterozygote
GM3178	2	HF12 20U	0023	30	F	WH	+-(O)		MOTHER	600	3	XPH1AA; Asian
GM3179	2	HF12 20U	0023	32	M	WH	+-(O)		FATHER	600	4	XPH2AA; Asian
Pigmented Xerodermoid												
*GM3379	2	HF12 20U	0323	86	F	WH	-?		PROBAND	220	1	X.P. variant; late onset; normal excision repair; prolonged post UV DNA synthesis; see GM3456 Lymphoid
DISORDERS OF STEROID METABOLISM												
Adrenal Hyperplasia (Adrenogenital Syndrome) Type III - 20191												
GM2241	2	HF12 20U	0107	1MO	F	WH	--		PROBAND	85	1	Genital tissue; see GM2242 Lymphoid; 21-hydroxylase deficiency; salt-losing type; responsive to hydrocortisone treatment
Adrenoleukodystrophy (Addison Disease and Cerebral Sclerosis) - 30010												
GM0337	3	MC5A 20U	0105	35	F	WH	+?		PARENT	86	2	Mother of an affected child; proband not in Repository
Pseudovaginal Perineoscrotal Hypospadias - 26460												
GM2935	9	HF12 20U	0077	16	F	WH	--		PROBAND	461	1	5-alpha-reductase deficiency; Arab; similarly affected sister
Testicular Feminization Syndrome - 31370												
GM1404	4	EEMEM 20U	0109	20	F	BL	y-		PROBAND			46,XY
GM2299	3	HF12 20U	0107	8MO	F	BL	y-		PROBAND	123	1	46,XY; genital skin fibroblast culture; half-sister of GM2682
GM2300	2	HF12 20U	0107	8MO	F	BL	y-		SAME	123	1	46,XY; non-genital skin fibroblast culture

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF STEROID METABOLISM											
Testicular Feminization Syndrome - 31370											
GM2301	4	HF12	20U 0107	8MO	F	BL	y-	SAME	123	1	46,XY; gonad fibroblast culture
GM2682	4	HF12	20U 0107	7	F	BL	y-	AFF SIS	123	2	Thigh skin; 46,XY
GM2683	4	HF12	20U 0107	7	F	BL	y-	AFF SIS	123	2	Genital skin; 46,XY; same patient as GM2682
GM2715	2	HF12	20U 0107	14	F	WH	y-	PROBAND	244	1	Genital skin; 46,XY
GM2716	3	HF12	20U 0107	14	F	WH	y-	SAME	244	1	Epididymus tissue; 46,XY
GM2717	2	HF12	20U 0107	14	F	WH	y-	SAME	244	1	Non-genital skin; 46,XY
GM1721	2	MC5A	20U 0107	15	F	WH	y-	PROBAND	393	1	46,XY; abdominal skin fibroblast culture; genital tissue fibroblast culture (GM1722) also available
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE											
Camptomelic Dwarfism - 21197											
GM1737	9	MC5A	20U 0142	9DA	F	WH	--	PROBAND			Gonad biopsy; phenotypically female; 46,XY with random chromosome loss
Cartilage-Hair hypoplasia (Metaphyseal Chondrodysplasia) - 25025											
GM1671	4	HF12	20U 0070	17	M		--	PROBAND			Slightly elevated level of adenosine deaminase activity in fibroblasts
GM3625	8	HF12	20U 0134	38	F		--	PROBAND			Amish; short stature; thin hair; no history of immune defect
GM3626	7	HF12	20U 0134	20	M	WH	--	PROBAND			Old order Amish; short stature, T cell defect, and thin hair
Chondrodystrophy											
GM0229	3	MC5A	20U 0107	9MO	M	WH	-?	PROBAND			Mother also affected; 46,XY
Cockayne Syndrome - 21640											
*GM0739	3	MC5A	20U 0115	3	F	WH	--	PROBAND			46,XX; increased UV light-induced sister chromatid exchanges and cell killing
GM1428	9	MC5A	20U 0091	10	F	BL	--	PROBAND			
GM2838	3	HF12	20U 0264	24	M	WH	--	PROBAND			Profound dwarfing; typical facies; ataxic; premature aging and death; two similarly affected sibs
*GM1098	4	EEMEM	20U 0006	20	M	WH	--	PROBAND	251	1	46,XY; see GM1712 Lymphoid; sensitive to UV irradiation

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DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Cockayne Syndrome - 21640												
*GM1856	4	HF12	20U 0155	13	M	WH	--	B	PROBAND	399	1	Fibroblasts have markedly decreased post U.V. light colony forming ability; see GM1857 Lymphoid
*GM1629	3	HF12	20U 0155	10	F	WH	--	B	PROBAND	628	1	Father is GM2474 Lymphoid; mildly retarded; small skull; spastic diplegia; fibroblasts have decreased post UV light colony forming ability
GM2965	2	HF12	20U 0210	25	F	WH	--		PROBAND	636	1	Atypical; late age of onset; lack of ocular and retinal lesions; see GM2964 Lymphoid
Conradi Syndrome (Chondrodysplasia Punctata) - 21510												
GM0740	2	MC5A	20U 0107	1	F	WH	--		PROBAND			
Cornelia de Lange Syndrome - 21790												
GM0045	5	MC5A	20U 0066	13	M	WH	--		PROBAND			46,XY; some chromosome loss and structural aberrations in 70% of fibroblasts
GM3478	2	HF12	20U 0107	4DA	M	WH	--		PROBAND			Typical facies; clinodactyly
Cutis Laxa - 21910												
GM0480		MC5A	20U 0066	45	F	WH	--		PROBAND			Passage 4 at IMR
GM1353	3	MC5A	20U 0147	12DA	M	WH	--		PROBAND			46,X,Yp+
GM1377	4	EEMEM	20U 0147	8WK	M	BL	--		PROBAND			Redundant skin on arms and thighs; hyperextensible MP and IP joints
GM2739	2	HF12	20U 0232	17	M	WH	--		PROBAND			Classical symptoms
GM2768	5	HF12	20U 0147	4MO	F	WH	--		PROBAND			
Dyschondrosteosis - 12730												
GM2885	2	HF12	20U 0315	47	F	WH	+-		PROBAND			Type of Leri and Weil
Dystonia Musculorum Deformans, Dominant Form - 12810												
GM3211	2	HF12	20U 0307	30	M	WH	+-		PROBAND			Generalized dystonia; Ashkenazi; age of onset 7 yrs; mother and sister also affected
GM3221	3	HF12	20U 0307	50	F	WH	+-		PROBAND			Focal dystonia; father and grandfather also affected; age of onset 43 yrs
GM2304	3	HF12	20U 0184	17	F	WH	+-		PROBAND	102	1	See GM2305 Lymphoid; onset at age 7; at time of biopsy very little speech and considerable dystonia of all extremities

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DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Dystonia Musculorum Deformans, Dominant Form - 12810												
GM2348	5	HF12	20U	0184	21	F	WH	+-	PROBAND	218	1	Swedish background; age of onset 5 yrs; see GM2347 Lymphoid
Dystonia Musculorum Deformans, Recessive Form - 22450												
GM3208	2	HF12	20U	0307	29	M	WH	--	PROBAND			Generalized dystonia; Ashkenazi; age of onset 10 yrs
GM3218	2	HF12	20U	0307	25	M	WH	--	PROBAND			Generalized dystonia; similarly affected brother; age of onset 2 yrs
GM2306	2	HF12	20U	0184	13	M	WH	--	PROBAND	103	1	See GM2307 Lymphoid; onset at age 10; Jewish background
GM2255	3	HF12	20U	0184	14	F		--	PROBAND	473	1	See GM2256 Lymphoid; onset at age 12; severe involvement of trunk and extremities; similarly affected sister
GM2551	3	HF12	20U	0184	31	F	WH	--	PROBAND	517	1	Eastern European Jewish; onset at age 15
Dystonia Musculorum Deformans: Type Unknown												
GM3209	3	HF12	20U	0307	49	F	WH		PROBAND			Focal (axial) dystonia; age of onset 41 yrs
GM3210	3	HF12	20U	0307	21	M	WH		PROBAND			Generalized dystonia; age of onset 2 yrs; negative family history
GM3213	2	HF12	20U	0307	26	F	WH		PROBAND			Generalized dystonia; age of onset 12 yrs
GM3214	3	HF12	20U	0307	37	M	WH		PROBAND			Focal (spasmodic torticollis) dystonia; age of onset 35 yrs; negative family history
GM3215	2	HF12	20U	0307	48	F	WH		PROBAND			Segmental dystonia; age of onset 44 yrs; negative family history
GM3216	2	HF12	20U	0307	64	F	WH		PROBAND			Focal (oral-mandibular) dystonia; age of onset 59 yrs; negative family history
GM3217	2	HF12	20U	0307	49	M	WH		PROBAND			Segmental dystonia; age of onset 41 yrs; negative family history
GM3219	4	HF12	20U	0307	18	M	WH		PROBAND			Segmental dystonia; Ashkenazi; age of onset 10 yrs; negative family history
GM3220	4	HF12	20U	0307	35	M	WH		PROBAND			Focal (spasmodic torticollis) dystonia; age of onset 33 yrs; negative family history

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Dystonia Musculorum Deformans: Type Unknown												
GM3222	4	HF12	20U 0307	71	F	WH			PROBAND			Focal (spasmodic torticollis) dystonia; Ashkenazi; age of onset 67 yrs; negative family history
Ehlers-Danlos Syndrome: Type I - 13000												
GM1812	3	HF12	20U 0171		F		+-		PROBAND			
GM2007	6	HF12	20U 0171	38	F	WH	+-		PROBAND			
GM2691	6	HF12	20U 0147	17	F	WH	+-		PROBAND			
Ehlers-Danlos Syndrome: Type II - 13000												
GM1788	2	HF12	20U 0171	51	M	WH	+-		PROBAND			
GM1691	3	HF12	20U 0171	19	M	WH	+-		PROBAND	104	1	Same patient as GM2006
Ehlers-Danlos Syndrome: Type IV - 13005, 22535												
GM2022	7	EEMEM	10U 0147	18	F	WH			PROBAND	362	1	Recurrent aneurysms; blue sclerae; hypomobile thumbs; mode of inheritance uncertain; negative family history
Ehlers-Danlos Syndrome: Type VI - (Hydroxylysine-deficient Collagen) - 22540												
*GM1790	15	EEMEM	20U 0147	12	F	WH	--		PROBAND	105	1	Parents not affected; kyphoscoliosis; recurrent joint dislocation; hyperextensible skin and joints; decreased collagen hydroxylysine content
*GM1791	15	EEMEM	20U 0147	9	F	WH	--		AFF SIS	105	2	Kyphoscoliosis; recurrent joint dislocation; hyperextensible skin and joints; decreased collagen hydroxylysine content
Ehlers-Danlos Syndrome: Type Unclassified												
GM0733	3	MC5A	20U 0143	7	M	WH			PROBAND			Physical diagnosis: affected
GM1161	3	EEMEM	20U 0095	8	F				PROBAND			Physical diagnosis: affected
GM1897	3	HF12	20U 0147	32	F	WH			PROBAND			Physical diagnosis: affected
GM2293	2	HF12	20U 0185	14	F	WH			PROBAND			Cuban; bilateral congenital glaucoma; loose skin; hyperpigmented skin; hypertrophic scars; father and brother are similarly affected
GM2681	2	HF12	20U 0147	36	F	WH			PROBAND			Physical diagnosis: affected
GM2206	3	HF12	20U 0147	17	F	WH		B	PROBAND	208	1	Variant; hydroxylysine deficient, not lysyl hydroxylase deficient; 46,XX,inv(6)(pter>p24::p21>p24::p21>qter)pat; formerly GM1514

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Ehlers-Danlos Syndrome: Type Unclassified												
GM2205	4	HF12	20U 0147	51	M	WH		A	FATHER	208	2	Clinically unaffected; 46,XY,inv(6)(pter>p24::p21>p24::p21>qter); formerly GM1515
GM1516	2	EEMEM	20U 0147	49	F	WH			MOTHER	208	3	Clinically unaffected; 46,XX
GM2490	9	EEMEM	10I 0147	11	F	WH			PROBAND	483	1	Joint laxity; thin skin; similarly affected sister
Epidermolysis Bullosa Dystrophica - 22660												
GM2857	2	HF12	20U 0232	12	M	WH	--		PROBAND			Puerto Rican; negative family history
Giant Mitochondrial Disease - 25540												
GM0028	4	MC5A	20U 0095	21	F	WH	--		PROBAND			Lebanese; 46,XX; skeletal muscle hypermetabolism; abnormal mitochondria; genetic etiology unproven; 2nd reported case
Hypophosphatasia - 24150												
GM3459	3	HF12	20U 0330	5MO	M	WH	--	B	PROBAND			Severe lethal, in utero onset; deficient serum and fibroblast alkaline phosphatase
*GM1571	3	MC5A	20U 0095	20FW	M	BL	--	B	PROBAND	371	1	Fetal; in utero onset; fatal; deficient fibroblast alkaline phosphatase activity; similarly affected sib
GM3191	3	HF12	20U 0078	30	M	WH	+-(0)		PARENT	509	1	Father of child with fatal, in utero onset form; proband not in Repository
GM3192	3	HF12	20U 0078	25	F	WH	+-(0)		PARENT	509	2	Mother of child with fatal, in utero onset form; proband not in Repository
GM3502		EEMEM	20U 0321	20FW	F	WH	--		PROBAND	604	1	See GM3336 Amniotic; severe, lethal, in utero onset; passage 2 at IMR; similarly affected sib
GM3328		EEMEM	20U 0321		F	WH	+-(0)		MOTHER	604	2	Passage 4 at IMR
GM3327		EEMEM	20U 0321	25	M	WH	+-(0)		FATHER	604	3	Passage 4 at IMR
Marfan Syndrome - 15470												
*GM0036	4	MC5A	20U 0026	10	M	WH	+-		PROBAND	112	1	46,XY; kyphoscoliosis; tall; long arms
*GM0035	4	MC5A	20U 0026	12	M	WH	+-		AFF BRO	112	2	46,XY; kyphoscoliosis; tall; long arms
Metaphyseal Dysplasia, Anetoderma & Optic Atrophy - 25045												
GM3129	3	HF12	20U 0232	27	F	WH	--		PROBAND	452	1	Uninvolved skin; macular anetoderma of Schweninger and Buzzi; Jewish

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DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Metaphyseal Dysplasia, Anetoderma & Optic Atrophy - 25045												
GM3130	2	HF12 20U	0232	27	F	WH	--		SAME	452	1	Involved skin
Winchester Disease: Mucopolysaccharidosis - 27795												
*GM2295	2	HF12 20U	0180	21	F	WH	--	B	PROBAND	477	1	Puerto Rican; rheumatoid arthritis; coarse facies; corneal opacity; increased intracellular uronic acid level in fibroblasts; similarly affected sis.
Muscular Dystrophy; Becker Type (Progressive Tardive) - 31010												
GM2298	8	EEMEM 15U	0202	18	M		y-		PROBAND			
Muscular Dystrophy; Duchenne Type (Pseudohypertrophic Progressive) - 31020												
GM2339	8	MC5A 20U	0088	20FW	M		y?		PROBAND			Presumptive affected fetus (50% risk); 46,XY
*GM3429	10	EEMEM 20U	0049	6	M	WH	y-		PROBAND			Can. Rep# 466; prog. proximal muscle weakness; elev. CPK; myopathic EMG; neg. family history
GM3604A	6	EEMEM 20U	0006	4	M	WH	y-		PROBAND			Abnormal EMG; abnormal muscle biopsy; elevated CPK; no family history of muscular dystrophies
GM3677	15	HF12 20U	0347	18	M	WH	y-		PROBAND			Upper & lower proximal muscle weakness; positive family hx; "hypertrophic" calf muscles; elev. CPK; not typical DMD
GM3781	3	EEMEM 20U	0006	11	M	WH	y-		PROBAND	533	1	Latin American; elevated serum CPK; see GM3780 Lymphoid
GM3783	2	EEMEM 20U	0006	10	M	WH	y-		AFF BRO	533	2	Latin American; elevated serum CPK; see GM3782 Lymphoid
Myositis Ossificans Progressiva - 13510												
GM0513	3	MC5A 20U	0008	16	M	WH	+-		PROBAND			
GM0783	3	MC5A 20U	0107		M	BL	+-		PROBAND			
Myotonic Dystrophy (Steinert Disease) - 16090												
GM3132	2	HF12 20U	0210	18	M	WH	+-		PROBAND			Bone deformities; muscle wasting; muscle myotonia
GM3755	3	HF12 20U	0348	44	M	WH	+-		PROBAND	488	1	Typical phenotype; mother, two brothers and a sister also affected; see GM3756 Lymphoid
GM3759	4	EEMEM 20U	0348	33	M	WH	+-		PROBAND	527	1	EMG myotonia; myotonic face; distal weakness and atrophy; positive family history; affected daughter is GM3696A Lymphoid

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE											
Osteogenesis Imperfecta - 16620											
GM1093	7	MC5A	20U 0095	8	M	WH	+-	PROBAND			
GM1436	3	MC5A	20U 0147	24	M	WH	+-	PROBAND			
GM1789	2	HF12	20U 0147	12	F	WH	+-	PROBAND			Scoliosis; slightly blue sclerae; negative family history
GM2573	3	HF12	20U 0147	17	M	WH	+-	PROBAND			
GM2645	2	HF12	20U 0147	17	M	WH	+-	PROBAND			Positive family history
GM2695	2	HF12	20U 0147	12	F	WH	+-	PROBAND			
Osteogenesis Imperfecta Congenita - 25940											
GM0744	6	EEMEM	20U 0057	1DA	F	WH	--	PROBAND			46,XX; negative family hx; multiple fractures of extremities, skull, & ribs present at birth; lethal
GM2328	2	HF12	20U 0107	4DA	M	WH	--	PROBAND			First affected child of clinically normal parents
GM2962	3	HF12	20U 0070	8DA	F	WH	--	PROBAND			"Broad boned"
GM3138	2	HF12	20U 0147	3	F	WH	--	PROBAND			Broken bones; opalescent teeth; blue sclera; poor motor development
Parana Hard-Skin Syndrome - 26053											
GM2663	2	HF12	20U 0147	13	M		--	PROBAND			
Pseudoachondroplastic Dysplasia IV - 26416											
GM3127	2	HF12	20U 0107	3	F		--	PROBAND			Sibling died of same disorder
Thanatophoric Dwarfism - 27365											
GM1422	2	MC5A	20U 0107	2WK	F	BL	--	PROBAND			Severely dwarfed; extremely short extremities; small hands and feet; large head; distended abdomen
Werdnig-Hoffmann Disease - 25330											
GM0232	2	MC5A	20U 0078	7MO	M	WH	--	PROBAND			Progressive muscular atrophy; abnormal EMG; absent deep tendon reflexes
OTHER DISORDERS OF KNOWN BIOCHEMISTRY											
Acatalasia (Acatalasemia) - 20020											
GM0064		EEMEM	12U 0080		M	WH	--	B PROBAND			Swiss; passage 5 at IMR; limited supply
GM0065		EEMEM	12U 0080		M	OR	--	B PROBAND			Japanese; passage 7 at IMR; limited supply

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAM#	FAMM#	REMARKS
OTHER DISORDERS OF KNOWN BIOCHEMISTRY												
Acatalasia (Acatalasemia) - 20020												
GM1931	8	MC5A	20U 0169	73	M	WH	+-(O)		PARENT			Swiss; father of an affected child
Porphyria: Hereditary Coproporphyria - 12130												
GM0962	3	EEMEM	20U 0112	59	F	WH	+-		PROBAND	641	1	46,XX; see GM2302 Lymphoid
Porphyria: Acute Intermittent Porphyria - 17600												
GM1647	3	HF12	20U 0112	44	F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase
*GM0933	3	HF12	20U 0112	15MO	F	WH	+-	A	DAU	81	1	Latent carrier; deficient URO synthetase
*GM0932	3	HF12	20U 0112	4	M	WH	+-	A	SON	81	2	Latent carrier; deficient URO synthetase
*GM0931	3	HF12	20U 0112	32	F	WH	+-	A	PROBAND	81	3	Active AIP; deficient URO synthetase
*GM0939	3	EEMEM	20U 0112	32	F	WH	+-	A	PROBAND	82	1	See GM2135 Lymphoid; active AIP; deficient URO synthetase
*GM0940	2	EEMEM	20U 0112	61	F	WH	++	A	MOTHER	82	2	See GM2134 Lymphoid; normal; no URO synthetase deficiency
*GM0941	3	EEMEM	20U 0112	63	M	WH	+-	A	FATHER	82	3	Latent carrier; see GM2133 Lymphoid; deficient URO synthetase
GM1623	2	HF12	20U 0112	16	M	WH	+-	A	AFF BRO	83	1	Deficient uroporphyrinogen-I synthetase
GM1622	3	HF12	20U 0112	19	F	WH	+-	A	PROBAND	83	2	Deficient uroporphyrinogen-I synthetase
*GM1621	2	HF12	20U 0112	39	F	WH	+-	A	AFF MOTHER	83	3	Deficient uroporphyrinogen-I synthetase
GM1625	3	HF12	20U 0112	3	M	WH	+-	A	AFF SON	84	1	Deficient uroporphyrinogen-I synthetase
GM1624	3	HF12	20U 0112	22	F	WH	+-	A	PROBAND	84	2	Deficient uroporphyrinogen-I synthetase
Porphyria: Cutanea Tarda - 17610												
GM0961	3	MC5A	20U 0112	70	M	WH	+-		PROBAND			
GM0977	5	HF12	20U 0112	81	F	WH	+-		PROBAND			
GM1041	6	HF12	20U 0112	69	F	WH	+-		PROBAND			
GM1082	2	HF12	20U 0112	21	M	WH	+-		PROBAND			
GM1179	3	HF12	20U 0112	36	M	WH	+-		PROBAND			
GM1482	4	HF12	20U 0112	58	M	WH	+-		PROBAND			
Sickle Cell Anemia (Hemoglobin S) - 14170												
GM2340	10	EEMEM	10I 0088	20FW	F		--		PROBAND			Sickle cell disease

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OTHER DISORDERS OF KNOWN BIOCHEMISTRY												
Thalassemias - 27350												
GM3037	5	EEMEM 20U	0088	23FW	M	OR	--	B	PROBAND			Alpha-thalassemia, fetal hydrops; Vietnamese; confirmed by cDNA hybridization and globin chain synthesis
GM3433	7	EEMEM 20U	0049	29FW	F	OR	--		PROBAND			Chinese; Canadian Rep. No. 646; electrophoresis confirmed Barts Hb as major component; alpha-thalassemia; positive family history
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Acrodermatitis Enteropathica - 20110												
GM2814	3	HF12 20U	0070	6MO	M	WH	--		PROBAND	323	1	Low serum zinc concentration
GM2815	3	HF12 20U	0070	24	M	WH	+-(O)		FATHER	323	2	
Alzheimer Disease of Brain - 10430												
GM0364	2	MC5A 20U	0139	53	M	WH	+-		PROBAND	87	1	Pedigree contains many affected individuals
GM0490	3	MC5A 20U	0139	33	F	WH	+?		NIECE	87	6	At risk
Antitrypsin Deficiency - 20740												
GM2522	2	HF12 20U	0107	3MO	M	WH	--		PROBAND			Serum Pi type is ZZ; both parents are MZ
Basal Cell Nevus Syndrome - 10940												
GM3300	2	HF12 20U	0107	11	F	WH	+-		PROBAND			Jaw cysts; cleft lip and palate; mental retardation; multiple nevi
GM1657	3	HF12 20U	0104	53	M	WH	+-		PROBAND	88	1	Normal skin; see GM1656 Lymphoid
GM1658	3	HF12 20U	0104	53	M	WH	+-		SAME	88	1	Basal cell carcinoma; see GM1656 Lymphoid
GM1725	3	HF12 20U	0104	58	F	WH	+-		PROBAND	89	1	See GM1726 Lymphoid
GM2098	3	HF12 20U	0104	31	M	WH	+-		PROBAND	90	1	See GM2099 Lymphoid
GM2138	4	HF12 20I	0104		F	WH	+-		PROBAND	91	1	See GM2139 Lymphoid
GM1577	5	HF12 20U	0104	39	M	WH	+-		PROBAND	372	1	See GM1578 Lymphoid
GM1575	2	HF12 20U	0104	31	F	WH	+-		PROBAND	373	1	See GM1576 Lymphoid
GM1552	2	HF12 20U	0104	27	M	WH	+-		PROBAND	637	1	See GM1553 Lymphoid
Beckwith-Wiedemann Syndrome (EMG Syndrome) - 22560												
GM0359	3	MC5A 20U	0107	8	M	WH	--		PROBAND	351	1	Negative family history

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Canavan Disease (Spongy Degeneration of the CNS) - 27190											
GM0059	2	MC5A	20U 0095	17MO	F		--	PROBAND	424	1	
GM0060	4	MC5A	20U 0095	2	M		--	AFF BRO	424	2	Brain tissue fibroblast culture
Chediak-Higashi Syndrome - 21450											
GM2075	3	HF12	20U 0126	18MO	F	WH	--	PROBAND			Photophobia; no nystagmus; cytoplasmic inclusions in WBC; partial oculocutaneous albinism
Coffin-Lowry Syndrome - 30360											
*GM2365	4	HF12	20U 0213	14	M	WH	y-	PROBAND			Mental retardation; short stature; unusual facies; tapering fingers; positive family history
GM3318	2	HF12	20U 0107	8	M	WH	y-	PROBAND	510	1	See GM3317 Lymphoid; mental retardation; coarse facies; positive family history
Cystic Fibrosis (Mucoviscidosis) - 21970											
*GM0142	3	MC5A	20U 0107	14	M	WH	--	PROBAND			Severe
GM0668	11	HF12	20U 0059	10	M	WH	--	PROBAND			Sweat chloride: 125 meq/L; also has cleft lip and palate
*GM0770	2	MC5A	20U 0060	19	M	WH	--	PROBAND			Classic pulmonary and pancreatic involvement
GM0997	8	HF12	20U 0059	10	M	WH	--	PROBAND			Diagnosed at age 7 mos; negative family history for CF; sweat chloride:120meq/L; on enzyme replacements
*GM0998	8	HF12	20U 0059	4	M	WH	--	PROBAND			Confirmed by dexamethasone resistance; sweat chloride: 126 meq/L; on enzyme therapy
GM0999	14	HF12	20U 0059	13	F	WH	--	PROBAND			Diagnosed at age 3; one affected and six normal sibs; sweat chloride: 144 meq/L; symptoms are GI and pulmonary
*GM1348	3	MC5A	20U 0107	18	F	WH	--	PROBAND			Severe; first diagnosed at age 7
GM1958	4	MC5A	20U 0178	13	M	WH	--	PROBAND			Diagnosed at age 3 mo.; similarly affected sib
GM2801	5	MC5A	20U 0060	15	M	WH	--	PROBAND			Sample from nasal polyp; pancreatic insufficiency; elevated sweat electrolytes
GM2828		MC5A	20U 0060	2?	F	WH	--	PROBAND			No pancreatic insufficiency; passage 4 at IMR

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
*GM1012	8	HF12	10U	0005	8	M	WH	--	PROBAND	92	1	
*GM1011	10	HF12	10U	0005	7	M	WH	--	AFF BRO	92	2	
GM1009	9	HF12	20U	0005	34	M	WH	+-(0)	FATHER	92	3	
GM1014	8	HF12	10U	0005	13	M	WH	--	PROBAND	93	1	
GM1013	9	HF12	20U	0005	11	M	WH	--	AFF BRO	93	2	
GM1708	12	HF12	20U	0005	33	M	WH	+-(0)	FATHER	93	3	Formerly GM1010
GM1957	6	MC5A	20U	0178	11	M	WH	--	PROBAND	94	1	Diagnosed at age 3 mo.; pancreatic insufficiency
GM1959	4	MC5A	20U	0178	10	M	WH	--	AFF BRO	94	2	
GM3465	3	HF12	20U	0107	4	M	WH	--	PROBAND	233	1	Pericardium biopsy; has two affected sibs; first diagnosed by sweat test at age 10 weeks
GM3466	2	HF12	20U	0107	4	M	WH	--	SAME	233	1	Skin biopsy; has two affected sibs; first diagnosed by sweat test at age 10 weeks
*GM1707	10	HF12	20U	0005	9	F	WH	--	PROBAND	253	1	Confirmed by dexamethasone resistance; formerly GM1008
GM2827	4	MC5A	20U	0060	23	F	WH	--	PROBAND	336	1	Pancreatic insufficiency and Diabetes Mellitus
GM2826	8	MC5A	20U	0060	51	F	WH	+-(0)	MOTHER	336	2	
GM2803	5	MC5A	20U	0060	27	M	WH	++	BRO	336	3	Unaffected
*GM0768	3	MC5A	20U	0060	13	M	WH	--	PROBAND	404	1	Mild; no pancreatic involvement; see GM0897 Lymphoid
GM0851	7	R1640	20U	0025	22	M	WH	--	PROBAND	426	1	See GM1531 Lymphoid
Diabetes Mellitus of the Pima Indian - 22210											
GM2557	3	HF12	20U	0215	29	F	AI		PROBAND			Both parents and 3 grandparents are diabetic; onset at age 28
GM2574	3	HF12	20U	0215	23	M	AI		PROBAND	230	1	Pima/Papago American Indian; onset at age 18; on Insulin; see GM2575 Lymphoid
GM2565	2	HF12	20U	0215	28	M	AI		PROBAND	231	1	Onset at age 27 yrs; not on Insulin; see GM2566 Lymphoid

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus of the Pima Indian - 22210											
*GM2416	3	HF12	20U	0215	42	F	AI	PROBAND	383	1	See GM2417 Lymphoid; mother and two sibs also diabetic
*GM2440	2	HF12	20U	0215	23	M	AI	PROBAND	384	1	Both parents are diabetic
GM2442	4	HF12	20U	0215	36	F	AI	PROBAND	385	1	Both parents are diabetic
GM2556	2	HF12	20U	0215	27	F	AI	PROBAND	386	1	Onset at age 26; obese; not on Insulin; mother also diabetic
Diabetes Mellitus: Maturity Onset Diabetes - 22210											
GM1237	3	EEMEM	20U	0034	30	M	WH	PROBAND	95	1	MODY; onset at age 12; see GM1247 Lymphoid
GM1219	5	MC5A	20U	0034	46	M	WH	AFF BRO	95	2	MODY; onset at age 29; see GM1246 Lymphoid
GM1122	3	EEMEM	20U	0034	33	F	WH	AFF SIS	95	3	MODY; onset at age 16; see GM1240 Lymphoid
GM1430	3	MC5A	20U	0034	35	M	WH	AFF BRO	95	4	MODY; onset at age 16; see GM1243 Lymphoid
GM1955	3	MC5A	20U	0034	44	M	WH	AFF BRO	95	6	MODY; onset at age 26; see GM1956 Lymphoid
GM1497	2	MC5A	20U	0034		F	WH	AFF SIS	95	7	MODY; see GM1498 Lymphoid
GM1435	5	MC5A	20U	0034	22	M	WH	AFF NEPH	95	8	MODY; onset at age 14; see GM1242 Lymphoid; son of GM1497
GM1496	3	MC5A	20U	0034	20	M	WH	AFF NEPH	95	9	MODY; onset at age 12; see GM1244 Lymphoid; son of GM1497
*GM1409	3	MC5A	20U	0034	15	F	WH	AFF NIECE	95	10	Maternal cousin of GM1435 & GM1496; only juvenile diabetic in family; onset at age 13; on Insulin; see GM1410 Lymphoid
GM1837	4	HF12	20U	0034	22	F	WH	AFF COU	95	11	MODY type II; onset at age 18; paternal 1st cousin once removed of the 6 sibs; see GM1838 Lymphoid
GM1486	3	MC5A	20U	0034	37	M	WH	PROBAND	291	1	Third generation of MOD; see GM1241 Lymphoid
Diabetes Mellitus: Juvenile Onset - 22210											
*GM0609	3	MC5A	20U	0100	15	F	WH	PROBAND			On Insulin

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus: Juvenile Onset - 22210											
*GM2637	4	R1640 20U	0095	17	M	WH		PROBAND	236	1	Onset at age 2; on Insulin; see GM2676 Lymphoid
GM2638	4	R1640 20U	0095	10	F	WH		AFF SIS	236	2	Onset at age 3; on Insulin; see GM2793A Lymphoid
GM2639	6	R1640 20U	0095	19	M	WH		BRO	236	3	Non-diabetic; see GM2675 Lymphoid
GM2640	4	R1640 20U	0095	41	M	WH		FATHER	236	4	Non-diabetic; see GM2741A Lymphoid; both parents had maturity onset diabetes
GM2641	5	R1640 10U	0095	41	F	WH		MOTHER	236	5	Non-diabetic; see GM3355 Lymphoid
GM2712	8	R1640 10U	0095	17	M	WH		PROBAND	243	1	Onset at age 14; on Insulin; see GM2713 Lymphoid
GM2704	9	R1640 10U	0095	45	M	WH		FATHER	243	2	Non-diabetic; see GM2705 Lymphoid
GM2706	11	R1640 10U	0095	45	F	WH		MOTHER	243	3	Non-diabetic; see GM2707 Lymphoid
GM2708	9	R1640 10U	0095	16	M	WH		BRO	243	4	Non-diabetic; see GM2709 Lymphoid
GM2710	12	R1640 10U	0095	10	M	WH		BRO	243	5	Non-diabetic; see GM2711 Lymphoid
GM2869	7	R1640 20U	0260	25	F	WH		SIS	243	6	Non-diabetic; see GM2728 Lymphoid
GM2868	7	R1640 20U	0260	24	M	WH		AFF BRO	243	7	Onset at age 22
GM2209	4	MC5A 10U	0095	31	F	WH		SIS	307	1	Non-diabetic; HLA type A1,B8; mother has Insulin controlled and father has diet controlled diabetes
GM2635		R1640 20U	0095	33	F	WH		PROBAND	307	2	Onset at age 10; on Insulin; HLA type A1,A2/B8,Bw15; passage 2 at IMR
GM3358	5	R1640 10U	0260	15	M	WH		PROBAND	606	1	Onset at age 6; on Insulin; see GM3235 Lymphoid
GM3359	5	R1640 10U	0260	13	M	WH		BRO	606	2	Non-diabetic; see GM3239 Lymphoid
GM3360	5	R1640 10U	0260	43	M	WH		AFF FATHER	606	3	Onset at age 14; on Insulin; see GM3236 Lymphoid
GM3361	6	R1640 10U	0260	17	M	WH		BRO	606	4	Non-diabetic; see GM3237 Lymphoid
GM3362	6	R1640 10U	0260	41	F	WH		MOTHER	606	5	Non-diabetic; see GM3238 Lymphoid
*GM1872	3	MC5A 20U	0095	12	F	WH		PROBAND	620	1	Onset at age 10; on Insulin; see GM2762 Lymphoid

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus: Juvenile Onset - 22210											
GM1873	3	MC5A	20U	0095	15	F	WH	AFF SIS	620	2	Onset at age 13; on Insulin; see GM2763 Lymphoid
GM1909	3	MC5A	20U	0095	19	M	WH	AFF BRO	620	3	Onset at age 4; on Insulin; hyper-tension; short stature; see GM2765 Lymphoid
GM1910	3	MC5A	20U	0095	21	F	WH	SIS	620	4	Non-diabetic; see GM3354A Lymphoid
GM1911	3	MC5A	20U	0095	20	M	WH	AFF BRO	620	5	Onset at age 4; on Insulin; see GM2764 Lymphoid
GM1875	4	MC5A	20U	0095	17	M	WH	BRO	620	6	Non-diabetic; see GM2866A Lymphoid
GM1874	4	MC5A	20U	0095	22	F	WH	SIS	620	7	Non-diabetic; mentally retarded; see GM2766 Lymphoid
GM1876	4	MC5A	20U	0095	18	M	WH	BRO	620	8	Non-diabetic; see GM2812A Lymphoid
GM1878	4	MC5A	20U	0095	42	F	WH	MOTHER	620	9	Non-diabetic; see GM2714 Lymphoid
GM1877	4	MC5A	20U	0095	50	M	WH	FATHER	620	10	Non-diabetic; see GM3356A Lymphoid
GM2613	8	R1640	10U	0095	24	F	WH	PROBAND	621	1	Onset at age 6; see GM2677 Lymphoid
GM2614	8	R1640	10U	0095	27	M	WH	AFF BRO	621	2	Onset at age 18 mo.; on Insulin; see see GM2678 Lymphoid
GM2615	8	R1640	10U	0095	29	M	WH	BRO	621	3	Non-diabetic; see GM2679 Lymphoid
GM2616	8	R1640	10U	0095	63	M	WH	FATHER	621	4	Non-diabetic; see GM2680 Lymphoid
GM2617	7	R1640	10U	0095	60	F	WH	MOTHER	621	5	Non-diabetic; see GM2785 Lymphoid
Diabetes Mellitus and Insipidus with Optic Atrophy - 22230											
GM1610	3	MC5A	20U	0095	13	F	WH	PROBAND	96	1	Onset at age 11; optic atrophy; on Insulin; see GM1795 Lymphoid
GM1609	7	MC5A	20U	0095	18	F	WH	AFF SIS	96	2	Onset at age 16; not on Insulin; optic atrophy; see GM1799 Lymphoid
GM1611	4	MC5A	20U	0095	15	F	WH	AFF SIS	96	3	Onset at age 13; optic atrophy; on Insulin
GM1701	4	HF12	20U	0095	42	M	WH	FATHER	96	4	Non-diabetic; see GM1797 Lymphoid
DiGeorge Syndrome - 18840											
GM3577	2	HF12	20U	0107	8MO	M	WH B	PROBAND			45,XY,t(3;22); absent thymus and para-thyroid glands; cleft lip and palate; congenital heart disease

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Dubowitz Syndrome - 22337												
GM2850	2	HF12	20U	0269	2	M	AI	--	PROBAND			46,XY, in lymphocytes; peculiar facies; short stature; sparse hair; mental retardation; negative family history
Dysautonomia (Riley-Day Syndrome) - 22390												
GM0732	2	MC5A	20U	0107	15MO	M	WH	--	PROBAND			Non-Jewish; more severe than classical form
GM0850	5	R1640	20U	0025	26	M	WH	--	PROBAND			
GM2343	6	EEMEM	10U	0191	24	F		--	PROBAND			Mild familial type
GM2341	7	EEMEM	10U	0191	17	M		--	PROBAND	98	1	Mild familial type
GM2342	6	EEMEM	10U	0191	19	M		--	AFF BRO	98	2	Mild familial type
Dyggve-Melchior-Clausen Disease - 22380												
GM2437	3	HF12	20U	0070		F		--	PROBAND			
Dyskeratosis Congenita - 30500												
*GM1774	2	HF12	20U	0107	7	M	WH	y-	PROBAND	99	1	See GM1775 Lymphoid; maternal grandfather was affected
GM1786	2	HF12	20U	0107	30	F	WH	+-(0)	MOTHER	99	2	
GM1787	2	HF12	20U	0107	78	F	WH	+?	MAT GR GRMR	99	3	Possible carrier
Eosinophilic Fascitis												
GM2870	2	HF12	20U	0293	64	M	WH		PROBAND			May synthesize abnormal collagen or mucopolysaccharide
Epidermodysplasia Verruciformis - 22640												
GM2984	11	HF12	20U	0248		M	WH	--	PROBAND			French; warts; squamous cell carcinoma of skin; negative family history
GM2972	10	HF12	20U	0011		M	WH	--	PROBAND	511	1	Erupted skin; higher than normal UV induced unscheduled DNA synthesis
GM2973	8	HF12	20U	0011		M	WH	--	SAME	511	1	Uninvolved skin
Fahr's Disease: Cerebral calcification, non-arteriosclerotic - 21360												
GM3242	2	HF12	20U	0078	3	F		--	PROBAND			46,XX; a sister is also affected; periventricular calcifications; abnormal EEG; developmental delay; glaucoma
Friedreich Ataxia - 22930+13660												
GM3665		EEMEM	15U	0344	13	F	WH		PROBAND			Limb and gait ataxia; scoliosis; proprioceptive sensory loss; 46,XX; negative family history; recessive inheritance; passage 3 at IMR

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Friedreich Ataxia - 22930+13660											
GM3295	3	HF12	20U 0210	63	M	WH		PROBAND	68	1	Son and father also affected; pes cavus; peripheral neuropathy; cerebellar ataxia; nystagmus; sensorineural hearing loss; possibly dominant inheritance
Gilles de la Tourette Syndrome - 13758											
GM2642	11	EEMEM	10U 0191	12	M		+-	PROBAND			No family history of disease; progressive movement disorder since age 4; symptoms include extraordinary and violent actions
GM2664	10	EEMEM	20U 0191	11	M		+-	PROBAND	523	1	Positive family history; abnormal body movements since age 3; formerly GM2643
Gout - 13890											
GM0432	3	MC5A	20U 0120	32	M	WH	+-	PROBAND			Severe gouty arthritis
Huntington's Disease - 14310											
GM1168	3	EEMEM	20U 0136	43	M	WH	+-	PROBAND			Slurred speech; upper extremity chorea; no family history available
*GM1169	2	EEMEM	20U 0136	50	M	WH	+-	PROBAND			Choreoathetotic movements of all extremities and slurred speech; positive family history of H.D.
*GM1187	3	MC5A	20U 0136	43	M	WH	+-	PROBAND			Slurred speech; left upper extremity dysmetria; unsteady gait; positive family history of H.D.
*GM1171	3	EEMEM	20U 0136	37	F	WH	+?	PROBAND	106	2	At risk (50%); a sister has H.D.
*GM2079	3	HF12	20U 0136	48	F	WH	+-	PROBAND	107	1	See GM2080 Lymphoid; father is also affected
*GM2077	7	HF12	20U 0136	25	F	WH	+?	DAU	107	2	At risk (50%); see GM2078 Lymphoid
GM2147	6	HF12	20U 0186	55	M	WH	+-	PROBAND	108	1	Mother was also affected; see GM2146 Lymphoid
GM2149	3	HF12	20U 0186	54	F	WH	++	SPOUSE	108	2	Normal; see GM2148 Lymphoid
GM2151	3	HF12	20U 0186	26	F	WH	+?	DAU	108	3	At risk (50%); see GM2150 Lymphoid; personality disorder but no chorea
GM2153	3	HF12	20U 0186	40	F	WH	++	SPOUSE	109	2	Normal widow of proband; see GM2152 Lymphoid
GM2155	3	HF12	20U 0186	20	F	WH	+?	DAU	109	3	At risk (50%); see GM2154 Lymphoid

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Huntington's Disease - 14310											
GM2157	4	HF12	20U	0186	16	F	WH	+?	DAU	109	4	At risk (50%); see GM2156 Lymphoid
GM2159	3	HF12	20U	0186	19	F	WH	+?	DAU	109	5	At risk (50%); see GM2158 Lymphoid
GM2161	3	HF12	20U	0186	21	M	WH	+?	SON	109	6	At risk (50%); see GM2160 Lymphoid
GM2163	3	HF12	20U	0186	12	M	WH	+?	SON	109	7	At risk (50%); see GM2162 Lymphoid
GM2165	2	HF12	20U	0186	55	M	WH	+-	PROBAND	110	1	Mother and sister were also affected; age of onset 46 years
GM2177	6	HF12	20U	0186	26	M	WH	+?	SON	110	2	At risk (50%); see GM2176 Lymphoid
GM2183	3	HF12	20U	0186	21	F	WH	+?	DAU	110	3	At risk (50%); see GM2182 Lymphoid
GM2169	3	HF12	20U	0186	52	M	WH	++	BRO IN LAW	110	4	Normal spouse of GM2166 Lymphoid (affected sister of proband); see also GM2168 Lymphoid
GM2187	3	HF12	20U	0186	60	F	WH	+?	SIS	110	5	Asymptomatic at age 60; reduced at risk; see GM2186 Lymphoid
GM2189	2	HF12	20U	0186	63	M	WH	++	BRO IN LAW	110	6	Normal spouse of GM2187; see GM2188 Lymphoid
GM2167A	5	EEMEM	20U	0186	61	F	WH	+-	AFF SIS	110	7	See GM2166 Lymphoid
GM3621	4	HF12	20U	0186	29	F	WH	+-	AFF DAU	110	10	See GM3620 Lymphoid; age of onset 18 yrs
GM2173	3	HF12	20U	0186	51	F	WH	+-	PROBAND	111	1	See GM2172 Lymphoid
GM2171	2	HF12	20U	0186	22	F	WH	+?	DAU	111	2	At risk (50%); see GM2170 Lymphoid
GM2175	2	HF12	20U	0186	55	M	WH	++	SPOUSE	111	3	See GM2174 Lymphoid; clinically normal
GM2123	2	HF12	20U	0190	7FW	U	WH	+?	PROBAND	206	1	Fetus at 25% risk for Huntington's Disease
GM2191	3	HF12	20U	0190	32	F	WH	+?	MOTHER	206	2	At risk (50%); see GM2190 Lymphoid
GM1136	4	EEMEM	20U	0136	45	M	WH	+-	PROBAND	286	1	
*GM0305	5	MC5A	20U	0305	56	F	WH	+-	PROBAND	288	1	Age at onset:46; uncertain family history; formerly GM0094; normal spouse is GM0288 Fibroblast
*GM1061	2	MC5A	20U	0136	51	M	WH	+-	PROBAND	292	1	Choreoathetosis; dysarthria; dysphagia; see GM3642 Lymphoid

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Huntington's Disease - 14310											
*GM1085	3	MC5A	20U 0136	44	M	WH	+-	PROBAND	293	1	Affected mother & grandmother; moderate ataxia, chorea, dysarthria, and dysphagia; see GM3644 Lymphoid
GM1083A	8	MC5A	20U 0136	47	F	WH	+-	AFF SIS	293	2	See GM3643 Lymphoid; choreoathetosis, dysarthria, and dysphagia; age of onset 35 yrs
Incontinentia Pigmenti - 30830											
*GM0492	4	MC5A	20U 0078	26	F		+-	PROBAND			Typical skin lesions appeared shortly after birth
GM1236	9	EEMEM	20U 0109	3	F	WH	+-	PROBAND			Increased chromosome breakage; 46,XX
Jeune Syndrome:Newborn Asphyxiating Thoracic Dystrophy - 20850											
GM3521	2	HF12	20U 0074	2	F	WH	--	PROBAND			46,XX; thoracic-pelvic-phalangeal dystrophy
Lipoatrophic Diabetes - 15165											
*GM1828	4	EEMEM	20U 0178	18	M	WH	+-	PROBAND			Original diagnosis was atypical progeria; line K.H.
Lowe Oculocerebrorenal Syndrome - 30900											
GM1676	2	HF12	20U 0107	10	M	WH	y-	PROBAND			Positive family history
GM3265	2	HF12	20U 0107	17FW	M	WH	y?	PROBAND	512	1	At risk (50%); 46,XY; a brother was affected; pedigree shows multiple affected individuals
Meckel Syndrome - 24900											
GM3206	2	HF12	20U 0107	NB	F	WH	--	PROBAND	640	1	Skin biopsy; occipital encephalocele; polycystic kidneys; microcephaly; cultures from muscle and pericardium also available
Mental Retardation, X-Linked - 30950											
GM1228	3	EEMEM	20U 0066	12	M	WH	y-	PROBAND	641	1	Positive family history; formerly GM0642
Neurofibromatosis (von Recklinghausen Disease) - 16220											
*GM0622	3	MC5A	20U 0095	8	M		+-	PROBAND			
*GM1633	3	HF12	20U 0104	61	M	WH	+-	PROBAND	113	1	Atypical; see GM1634 Lymphoid
GM1639	3	EEMEM	20U 0104	19	F	BL	+-	PROBAND	114	1	Normal skin; other fibroblasts also available; see GM1641 Lymphoid
GM1860	3	HF12	20U 0104	41	M	WH	+-	PROBAND	115	1	Normal skin; other fibroblasts also available; see also GM1861 Lymphoid

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Neuropathy, Congenital Sensory, with Anhidrosis - 25680											
GM2958	2	HF12	20U 0105	13	M	WH	--	PROBAND			Insensitivity to pain
Olivopontocerebellar Atrophy - 16440, 50, 60, 70, and 25830											
GM3561	3	HF12	20U 0110	22	F	BL		PROBAND	240	1	Probably type III; see GM3562 Lymphoid; positive family history
Papular Mucinosis (genetic or acquired)											
GM0948	3	EEMEM	20U 0124	60	M	WH		PROBAND	116	1	Non-lesioned gingiva
GM0950	2	EEMEM	20U 0124	60	M	WH		SAME	116	1	Non-lesioned leg skin
GM0951	3	EEMEM	20U 0124	60	M	WH		SAME	116	1	Lesioned leg skin
Polyposis, Intestinal: Type I (Familial Polyposis of the Colon) - 17510											
GM2355	2	HF12	20U 0212	13	F	WH	+-	PROBAND	479	1	Affected father, grandmother, great-grandfather, and greatgreatgrandmother
Polyposis, Intestinal: Type III (Gardner Syndrome) - 17530											
GM2694	7	HF12	20U 0225	40	F	WH	+-	PROBAND	525	1	Tubulovillus adenoma; polyps
GM3223	2	HF12	20U 0308	25	M	WH	+-	AFF SON	603	1	See GM3224 Lymphoid
GM3314	2	HF12	20U 0225	48	F	WH	+-	PROBAND	603	2	English; adenocarcinoma of colon; see GM3315A Lymphoid
Progeria-26410											
GM0989B	15	EEMEM	20U 0055	20	M		--	PROBAND			Atypical progeria with unspecified cachectic dwarfism
*GM0990	8	EEMEM	20U 0055		M		--	PROBAND			Atypical progeria; also hypertriglyceridemia and abnormal glucose tolerance test
GM0991A	9	EEMEM	20U 0055	4	M		--	PROBAND			Classical progeria
GM1177	9	EEMEM	20U 0049	9	M		--	PROBAND			From Canadian Repository; 46,XY
GM1178	18	EEMEM	20U 0049	34	M		--	PROBAND			From Canadian Repository; slow growing culture; patient died at age 34; small eyes; micrognathia; nystagmus; large tongue; cardiac murmur
*GM1972	8	HF12	20U 0091	14	F	WH	--	PROBAND			Classical progeria; 46,XX
Prader - Willi Syndrome - 26400											
GM3340	10	HF12	20U 0078	28	M	WH	--	PROBAND			46,XY/47,XY,+ small metacentric (G-size) chromosome; hypotonia; hypogonadism; hypomentia; obesity; short stature

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Retinoblastoma, Sporadic											
GM0913	3	EEMEM	20U 0065	2	M	WH	+?	PROBAND	117	1	Unilateral; tumor tissue fibroblast culture
GM0914	3	EEMEM	20U 0065	2	M	WH	+?	SAME	117	1	Conjunctiva fibroblast culture; 46,XY, 9qh+
Schizophrenia and Psychiatric Disorders - 18150											
GM1792	3	HF12	20U 0137	26	M	WH		PROBAND	119	2	See GM1793 Lymphoid; episodes of agitation, delusions of persecution, fear of assassination; father also affected
GM1835	3	HF12	20U 0137	27	F	WH		AFF SIS	119	3	See GM1836 Lymphoid; hospitalized; drugs; father also affected
GM2497	3	HF12	20U 0137	23	M	WH		PROBAND	176	1	See GM1488 Lymphoid; paralogical thinking, affective shielding, splitting of affect from content, suspiciousness
GM2503	2	HF12	20U 0137	27	F	WH		AFF SIS	176	4	See GM1490 Lymphoid; anorexia nervosa since adolescence; more schizoid than depressed
GM2038	2	HF12	20U 0137	22	M	WH		PROBAND	204	1	Skin biopsy; onset of symptoms at age 6; suicide
GM2039	3	HF12	20U 0137	22	M	WH		SAME	204	1	Liver biopsy fibroblast culture
GM2790	5	HF12	20U 0137	64	M			PROBAND	276	1	Primary affective disease, depressed; stuporous for months; serious depression
Smith-Lemli-Opitz Syndrome - 27040											
GM3044	3	MC5A	20U 0148	2WK	A	WH	--	PROBAND			45,XY,t(13;14)(13qter>cen>14qter) in fibroblasts
Spinocerebellar Ataxia											
GM1960	5	HF12	20U 0105	15	F	WH		PROBAND			Similarly affected brother; culture grows slowly
Telangiectasia, Hereditary Hemorrhagic - 18730											
GM3418	2	HF12	20U 0210	52	M	WH	+-	AFF PAT GRFR	211	1	Gastrointestinal bleeding; mucous telangiectasias
GM3419	2	HF12	20U 0210	7	F	WH	+-	AFF DAU	211	2	Mucocutaneous telangiectasias
GM3015	3	HF12	20U 0210	28	M	WH	+-	PROBAND	211	3	Rendu-Osler-Weber Disease; seizures; cutaneous, ocular, and gastric telangiectasias; father was also affected

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Tuberous Sclerosis - 19110											
GM1635	3	HF12	20U 0104	17	M	BL	+-	PROBAND	124	1	Shagreen patch; see GM1636 Lymphoid
GM1643	3	HF12	20U 0104	17	M	BL	+-	SAME	124	1	Normal skin; see also GM1636 Lymphoid
*GM1644	2	MC5A	20U 0104	20	F	BL	+-	PROBAND	125	1	Normal skin; other fibroblasts also available; see also GM1638 Lymphoid
GM2332	2	HF12	20U 0210	23	M	WH	+-	PROBAND	216	1	Normal skin; seizures; mental retardation; adenoma sebaceum; shagreen patches
GM2333	4	HF12	20U 0210	23	M	WH	+-	SAME	216	1	Subungual fibroma fibroblast culture
Wiskott-Aldrich Syndrome - 30100											
*GM1598	2	MC5A	20U 0107	4	M	WH	y-	PROBAND			No family history of the disease
Zellweger Syndrome - 21410											
GM0228	3	MC5A	20U 0107	2MO	M	WH	--	PROBAND			
X CHROMOSOME MARKERS											
Agammaglobulinemia - 30030											
*GM0362	3	MC5A	20U 0125	21	M	WH	y-	PROBAND			46,XY; normal RBC G6PD level, Xg(a) antigen positive
GM3654	7	HF12	20U 0352	26	M	WH	y-	PROBAND			Russian-Italian; infantile, Bruton-type agammaglobulinemia; three affected generations
Glucose-6-Phosphate Dehydrogenase Deficiency & Variants - 30590											
*GM0120	9	EEMEM	20U 0041	30	F	BL	B	PROBAND			G6PD-A,PGK-1/G6PD-B,PGK-2
GM0324	5	MC5A	20U 0125	22	M	WH		PROBAND			Sardinian; homozygous for G6PD (med. type), Xg(a) antigen present
GM0325	4	MC5A	20U 0125	30	M	WH		PROBAND			Sardinian; heterozygous for G6PD deficiency (med. type), Xg(a) antigen present
GM0412	3	MC5A	20U 0009	19	M	WH		PROBAND			G6PD deficiency; Panama type
*GM0738	7	MC5A	20U 0041		M	WH		PROBAND			Hektoen variant of G6PD
GM1165	2	EEMEM	20U 0125	36	F	WH		PROBAND			46,XX; double heterozygote G6PD(B) Xg(a)-/G6PD(med)Xg(a)+
GM1166	3	EEMEM	20U 0125	39	F	WH		PROBAND			46,XX; double homozygote, G6PD(B) Xg(a)-/ G6PD(B) Xg(a)-; 0.75 units G6PD in fibroblasts

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X CHROMOSOME MARKERS											
Glucose-6-Phosphate Dehydrogenase Deficiency & Variants - 30590											
GM3165	2	HF12	20U 0117	8	M	WH		PROBAND			Hemolysis; RBC G6PD deficient; Greek; 0.15 units G6PD in fibroblasts
GM3171	2	HF12	20U 0117	16	M	WH		PROBAND			Hemolysis; RBC G6PD deficient; Greek; 0.10 units G6PD in fibroblasts
GM3172	3	HF12	20U 0117	39	F	WH		PROBAND			Favism; hemolysis; RBC G6PD deficient; Greek
GM3173	4	HF12	20U 0117	43	M	WH		PROBAND			Absent red cell G6PD; Greek; 0.14 units G6PD in fibroblasts
GM3174	3	HF12	20U 0117	40	F	WH		PROBAND			Absent red cell G6PD; Greek; 0.13 units G6PD in fibroblasts
GM3175	2	HF12	20U 0117	36	M	WH		PROBAND			Hemolysis; RBC G6PD deficient; 0.11 units G6PD in fibroblasts; Greek
GM1150	4	EEMEM	20U 0125	23	M	WH		PROBAND	157	1	46,XY; normal RBC G6PD(B) level, Xg(a) antigen absent; Sardinian
GM1152	4	EEMEM	20U 0125	49	M	WH		FATHER	157	2	46,XY; RBC deficient for G6PD(med type), Xg(a) antigen positive; Sardinian
GM1163	4	EEMEM	20U 0125	15	M	WH		BRO	157	3	46,XY; deficient RBC G6PD, Xg(a) antigen positive; Sardinian
GM1871	3	HF12	20U 0125	47	F	WH		MOTHER	157	4	46,XX; double heterozygote; G6PD(B) Xg(a)+/G6PD(med type)Xg(a)-
GM1151	3	EEMEM	20U 0125	12	M	WH		PROBAND	158	1	46,XY; RBC deficient for G6PD(med type), Xg(a) antigen absent; Sardinian
GM1154	4	EEMEM	20U 0125	37	F	WH		MOTHER	158	2	46,XX; double heterozygote; G6PD(med type)Xg(a)-/G6PD(B)Xg(a)+; Sardinian
GM1153	4	EEMEM	20U 0125	42	M	WH		FATHER	158	3	46,XY; normal RBC G6PD level, Xg(a) antigen positive; Sardinian; 0.53 units G6PD in fibroblasts
GM1188	3	MC5A	20U 0125		M	WH		PROBAND	159	1	46, XY G6PD (Seattle), Xg(a) antigen positive; Sardinian
GM1157	5	EEMEM	20U 0125	42	F	WH		MOTHER	159	2	46,XX; double heterozygote G6PD(Seattle) Xg(a)+/G6PD(B)Xg(a)-; Sardinian
GM1193	4	MC5A	20U 0125	50	F	WH		PROBAND	160	1	46,XX; RBC are heterozygous G6PD(B)/ G6PD(med) but are lacking enzyme activity; Sardinian

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			X CHROMOSOME MARKERS									
Glucose-6-Phosphate Dehydrogenase Deficiency & Variants - 30590												
GM1192A	7	MC5A	20U 0125	70	F	WH			MOTHER	160	2	46,XX; RBC are heterozygous G6PD(B)/ G6PD(med) but have normal levels of enzyme activity; Sardinian
GM2334	4	HF12	20U 0125		F	WH			MOTHER	217	2	46,XX; heterozygote; G6PD(B)/G6PD(med), Xg(a) antigen positive; son is GM2335 Fibroblast
GM2336	3	HF12	20U 0125		M	WH			FATHER	217	3	46,XY; G6PD(B), Xg(a) antigen positive; son is GM2335 Fibroblast
GM1886	3	HF12	20U 0125	11	M	WH			PROBAND	290	1	46,XY; G6PD(med), Xg(a) antigen absent
GM1887	3	HF12	20U 0125	7	M	WH			BRO	290	2	46,XY; G6PD(B), Xg(a) antigen absent; 0.76 units G6PD in fibroblasts
GM1888	2	HF12	20U 0125	14	M	WH			BRO	290	3	46,XY; G6PD(B), Xg(a) antigen absent
*GM2026	4	HF12	20U 0125	32	F	WH			PROBAND	465	2	46,XX; heterozygote; G6PD(med)/G6PD(B), Xg(a) antigen present
GM1869	2	HF12	20U 0125	15	M	WH			PROBAND	504	1	46,XY; normal RBC G6PD(B) level, Xg(a) antigen absent; 0.69 units G6PD in fibroblasts
GM1870	2	HF12	20U 0125	14	M	WH			MAT 1 COUS	504	2	46,XY; RBC deficient for G6PD(med), Xg(a) antigen absent
Ichthyosis: X-Linked - 30810												
GM3034	2	HF12	20U 0125	10	M	WH	y-	A	PROBAND			Sterol sulfatase deficiency; G6PD(med), Xg(a) antigen absent
GM3273	3	HF12	20U 0125	34	M	WH	y-	B	PROBAND			Sardinian; G6PD normal, Xg(a) negative; Sterol sulfatase deficient
GM3276	3	HF12	20U 0125	13	M	WH	y-	B	PROBAND			Sardinian; G6PD deficient, Xg(a) positive; Sterol sulfatase deficient
GM3277	2	HF12	20U 0125	15	M	WH	y-	B	PROBAND			Sardinian; G6PD normal, Xg(a) positive; Sterol sulfatase deficient
GM3278	3	HF12	20U 0125	26	M	WH	y-	B	PROBAND			Sardinian; G6PD normal, Xg(a) positive; Sterol sulfatase deficient
GM3280	4	HF12	20U 0125	46	M	WH	y?	B	PROBAND			Sardinian; G6PD normal, Xg(a) positive; Sterol sulfatase normal; possible variant

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMN#	REMARKS
X CHROMOSOME MARKERS												
Ichthyosis: X-Linked - 30810												
GM3281	3	HF12	20U 0125	48	F	WH	+?	B	PROBAND			Sardinian; G6PD(med), Xg(a)-/ G6PD(B), Xg(a)+; Sterol sulfatase normal
GM3283	3	HF12	20U 0125	37	F	WH	+?	B	PROBAND			Sardinian; G6PD(B), Xg(a)-/G6PD(med), Xg(a)+; Sterol sulfatase normal
GM3289	3	HF12	20U 0125	27	M	WH	y-	B	PROBAND			Sardinian; G6PD normal, Xg(a) positive; Sterol sulfatase deficient
GM3291	3	HF12	20U 0125	60	M	WH	y-	B	PROBAND			Sardinian; G6PD normal, Xg(a) positive; Sterol sulfatase deficient
GM3030	2	HF12	20U 0125	9	M	WH	y-	A	PROBAND	434	1	Sterol sulfatase deficiency; normal RBC G6PD, Xg(a) antigen positive
GM3031	5	HF12	20U 0125	47	F	WH	+?	A	MOTHER	434	2	G6PD (B)/G6PD (med); normal Sterol sulfatase
GM3033	5	HF12	20U 0125	66	F	WH	+-(0)	A	PARENT	435	2	G6PD(B)/G6PD(med), normal Sterol sulfatase; mother of 3 affected sons
GM3035	4	HF12	20U 0125	20	M	WH	y-	A	PROBAND	436	1	Sterol sulfatase deficiency; G6PD(med), Xg(a) antigen absent
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE												
Ataxia-Telangiectasia - 20890												
*GM0367	3	MC5A	20U 0107	17	M	WH	--		PROBAND			Fibroblast culture from lung biopsy; a deceased sister had A.T.
GM0647	7	EEMEM	20U 0133	17	M	WH	--		PROBAND			Neurologic disability and dramatic telangiectasia; elevated serum alpha-feto protein
*GM0648	9	EEMEM	20U 0133	15	M	WH	--		PROBAND			Neurologic disability and dramatic telangiectasia; abnormal chromosomes, polyploidy
*GM1588	13	HF12	20U 0077		M	WH	--		PROBAND			46,XY
GM1829	10	EEMEM	20U 0133	7	M	WH	--		PROBAND			
GM1841	11	EEMEM	20U 0133	13	M		--		PROBAND			
GM2052	3	HF12	20U 0077	15	F	WH	--		PROBAND			46,XX; Jewish
GM2797	4	EEMEM	20U 0109	40	F	WH	+-(0)		PARENT			Amish; mother of affected child

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE											
Ataxia-Telangiectasia - 20890											
GM1936	6	HF12 10U	0077	16	F	WH	--	PROBAND	126	1	
GM1937	7	HF12 10U	0077	24	F	WH	--	AFF SIS	126	2	
GM2531	6	HF12 20U	0077	5	M	WH	--	PROBAND	228	1	Iranian; see GM2782 Lymphoid
GM2530	7	HF12 20U	0077	8	M	WH	--	AFF BRO	228	2	Iranian; see GM2783 Lymphoid
*GM1740	18	HF12 20U	0077		M	WH	--	PROBAND	419	1	Moroccan; a deceased sister had A.T.; same patient as GM1939
GM3488	3	HF12 20U	0226	41	M	WH	+-(O)	FATHER	516	1	Clinically normal; see GM3187 Lymphoid
GM3489	2	HF12 20U	0226	37	F	WH	+-(O)	MOTHER	516	2	Clinically normal; see GM3188 Lymphoid
GM3487	3	HF12 20U	0226	8	M	WH	--	PROBAND	516	3	See GM3189 Lymphoid; neurological abnormalities
GM3490	3	HF12 20U	0226	16	F	WH	+?	SIS	516	4	See GM3324 Lymphoid; possible heterozygote
GM3492	2	HF12 20U	0226	7	M	WH	+?	BRO	516	5	Clinically normal; see GM3323 Lymphoid; possible heterozygote
GM3491	2	HF12 20U	0226	15	F	WH	+?	SIS	516	6	See GM3325 Lymphoid; possible heterozygote
GM3395	3	HF12 20U	0226	13	M	BL	--	PROBAND	605	1	See GM3332 Lymphoid; a deceased sister had A.T.
GM3397	2	HF12 20U	0226	43	M	BL	+-(O)	FATHER	605	2	See GM3382 Lymphoid
GM3396	2	HF12 20U	0226	42	F	BL	+-(O)	MOTHER	605	3	See GM3334 Lymphoid
GM3400	2	HF12 20U	0226	11	F	BL	+?	SIS	605	4	Normal; possible heterozygote; see GM3333 Lymphoid
GM3399	3	HF12 20U	0226	20	F	BL	+?	SIS	605	5	Normal; possible heterozygote; see GM3380 Lymphoid
GM3398	2	HF12 20U	0226	22	M	BL	+?	BRO	605	6	Normal; possible heterozygote; see GM3383 Lymphoid
Bloom Syndrome - 21090											
GM1492B	9	HF12 20U	0077	15	M	WH	--	PROBAND			B.S. Registry #44 (AbRu); Ashkenazi
*GM1493C	10	HF12 20U	0077	4	M	WH	--	PROBAND			B.S. Registry #57 (AmEl); Ashkenazi

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE											
Bloom Syndrome - 21090											
*GM2520	6	HF12	20U 0178	10	F	WH	--	A PROBAND			B.S. Registry # 20(ViSh); 46,XX; increased frequency of sister chromatid exchange
*GM2548	3	HF12	20U 0070	6	M	BL	--	A PROBAND			B.S. Registry# 71(HaEn); American; 46,XY; increased frequency of sister chromatid exchange
GM3498	7	EEMEM	20U 0134	3	M	WH	--	B PROBAND			B.S. Registry #87(AlFr); 46,XY; excessive sister chromatid exchanges
*GM3509	13	EEMEM	20U 0044	3	M	WH	--	PROBAND			B.S. Registry #81 (MaGrou), line HG1219; French Canadian; increased breakage and sister chromatid exchange
*GM3510	8	EEMEM	20U 0044	20	M	WH	--	B PROBAND			B.S. Registry #47(ArSmi), line HG1251; Ashkenazi; increased breakage and sister chromatid exchange
*GM3402	3	HF12	20U 0110	30	M	WH	--	PROBAND	210	1	Ashkenazi; B.S. Registry #9(EmSh); congenital dwarf; photosensitive telangiectatic eruption of the face
*GM1620	19	EEMEM	20U 0044	18MO	F	WH	--	PROBAND	430	1	B.S. Registry #53(StAs); Ashkenazi; Line HG916
*GM0811	17	EEMEM	10I 0044	4	M	WH	--	A PROBAND	500	1	B.S. Registry# 3(HoCo); Ashkenazi
*GM2932	8	HF12	20U 0077	28	M	WH	--	SAME	500	1	B.S. Registry# 3(HoCo); Ashkenazi
Fanconi Anemia (Pancytopenia) - 22765											
*GM0368	6	EEMEM	10U 0133	8	M	BL	--	A PROBAND			Multiple chromosome breaks; 46,XY
*GM0369	11	EEMEM	10U 0133	6	M	WH	--	PROBAND			Multiple chromosome breaks and gaps
*GM0391	6	EEMEM	10U 0133	6	F	WH	--	PROBAND			Multiple chromosome breaks
*GM0449	10	EEMEM	10I 0044	6	F	WH	--	PROBAND			46,XX; classical symptoms
*GM1309	3	EEMEM	20U 0133	12	M	BL	--	PROBAND			Lymphocytes showed multiple chromosome breaks
GM1746	5	MC5A	20U 0077	15	M	WH	--	PROBAND			Arab; 46,XY in leukocytes with quadri and tri radials and breaks
*GM2053	4	HF12	20U 0077	12	F	WH	--	B PROBAND			46,XX; multiple breaks, gaps, occasional dicentrics; Arab

HUMAN FIBROBLAST CULTURES WITH BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE												
Fanconi Anemia (Pancytopenia) - 22765												
GM2061	8	MC5A 20U	0133	20	F	WH	--		PROBAND			46,XX in leukocytes with chromosome breaks; several affected sibs
*GM2361	8	EEMEM 15U	0188	14	M		--		PROBAND			
GM2364	12	EEMEM 15U	0188	24	M		--		PROBAND			
GM0646	7	EEMEM 20U	0133	16	F	WH	--		PROBAND	365	1	
GM2977	9	EEMEM 20U	0133	52	M	WH	+-(0)		FATHER	365	2	
GM2975	9	EEMEM 20U	0133	40	M	WH	+-(0)		PARENT	464	1	Father of affected child; proband absent
GM2976	9	EEMEM 20U	0133	43	F	WH	+-(0)		PARENT	464	2	Mother of affected child; proband absent
GM2362	11	EEMEM 15U	0188	14	M		--		PROBAND	505	1	
GM2363	10	EEMEM 15U	0188	15	M		--		AFF BRO	505	2	
GONADAL DYSGENESIS												
XX Male												
GM1889	3	HF12 20U	0125	24	M	WH			PROBAND			46,XX; phenotypically male; G6PDB,Xg(a) antigen positive
*GM2626	9	R1640 20U	0030	23	M	WH		B	PROBAND	234	1	46,XX; second cousin of GM2670; bilateral mild gynecomastia; average I.Q.
*GM2624	12	R1640 20U	0030	45	M	WH			FATHER	234	2	46,XY in leukocytes
*GM2625	9	R1640 20U	0030	43	F	WH			MOTHER	234	3	46,XX in leukocytes
*GM2670	7	R1640 15U	0030	19	M	WH		B	PROBAND	235	1	46,XX; second cousin of GM2626; bilateral gynecomastia
*GM2671	7	R1640 15U	0030	47	F	WH			MOTHER	235	2	46,XX in leukocytes; clinically normal
*GM2672	11	R1640 20U	0030	48	M	WH			FATHER	235	3	Transient gynecomastia at age 20; in leukocytes, 2 cells out of 50 showed 46,XY,inv(7)(p15q36), remainder were normal
XY Female - 30610												
GM2600	5	EEMEM 20U	0109	31	F	WH			PROBAND			46,XY, H-Y antigen positive
GM2601	6	EEMEM 20U	0109	28	F				PROBAND			46,XY, H-Y antigen positive; male pseudohermaphroditism

HUMAN FIBROBLAST CULTURES WITH
BIOCHEMICAL MUTANT CONDITIONS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
GONADAL DYSGENESIS											
XY Female - 30610											
GM1628	2	MC5A	20U	0125	1	F	WH	A	PROBAND	122	1	46,XY; H-Y antigen positive; G6PDB, Xg(a) antigen positive
GM1948	3	HF12	20U	0125	27	F	WH		MOTHER	122	2	Normal
GM0048	7	MC5A	20U	0095	1	F	WH		PROBAND	156	1	46,XY; right gonadal tissue, H-Y antigen ?
GM0083	7	MC5A	20U	0095	1	F	WH	B	SAME	156	1	46,XY; left gonadal tissue, H-Y antigen?
GM3368	4	MC5A	20U	0172	17	F	BL	B	PROBAND	513	1	Pure gonadal dysgenesis; 46,XY H-Y antigen ?; right gonadal tissue
GM3368A	6	MC5A	20U	0172	17	F	BL	B	SAME	513	1	Pure gonadal dysgenesis; 46,XY H-Y antigen ?; left gonadal streak

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
TRANSLOCATION												
CHROMOSOME X												
*GM0089	5	MC5A	20U 0043		F	WH	46,X,t(X;19)(Xpter>Xq2::19q13 or 19p13> 19qter or 19pter;19pter or 19qter> 19p13 or 19q13::Xq2>Xqter)	B	A PROBAND			
*GM0097	3	MC5A	20U 0107	1	F	WH	46,X,t(X;1)(Xpter>Xq26::1q12>1qter; 1pter>1q12::Xq26>Xqter)	B	A PROBAND			De novo translocation; Beckwith-Wiedemann syndrome
*GM0705	13	HF12	20U 0016	10MO	F	WH	46,X,t(X;9)(Xpter>Xq12::9p24>9pter; 9qter>9p24::Xq12>Xqter)	B	A PROBAND			Culture grows slowly
*GM1695	5	HF12	20U 0176	26	F	WH	46,X,t(X;11)(Xqter>Xp2105::11q13>11qter; 11pter>11q13::Xp2105>Xpter)	B	A PROBAND			Also has clinically documented Duchenne Muscular Dystrophy
*GM1696	9	EEMEM	20U 0176	30	F	WH	46,X,t(X;7)(Xpter>Xq21::7p22>7pter; Xqter>Xq21::7p22>7qter)	B	A PROBAND			
GM1813	3	HF12	20U 0066	19	F	WH	46,XX,t(1;21)(q12;q22)pat/46,XX,t(1;21) (q12;q22)pat,t(X;15)(p11;q13)	U	A PROBAND			Menstrual dysfunction; Adj-2 segregation; Mosaic 40%/60%
GM2103	6	EEMEM	20U 0066	19	F	WH	46,X,t(X;Y)(Xpter>Xq11::Yq11>Yqter)	U	A PROBAND			Secondary amenorrhea
GM2469	2	HF12	20U 0070	17	F		46,X,t(X;Y)(Xpter>Xq22::Yq11>Yqter)	U	A PROBAND			
*GM2621	8	EEMEM	10U 0224	32	F	WH	46,X,t(X;12)(Xpter>Xq22::12q24>12qter; 12pter>12q24::Xq22>Xqter)	B	A PROBAND			Gonadal dysgenesis
*GM3322	4	HF12	20U 0070	35	F	WH	46,X,t(X;11)(Xpter>Xq22::11q13>11qter; 11pter>11q13::Xq22>Xqter)	B	A PROBAND			
GM3552	7	EEMEM	20U 0096	29	F	OR	46,X,t(X;11)(Xpter>Xq26::11q23> 11qter;11pter>11q23::Xq26>Xqter)	B	A PROBAND			Vietnamese; gonadal dysgenesis
*GM1414	5	EEMEM	20U 0068	28	F	BL	46,X,+der(9),t(X;9)(9pter>9q34::Xq13> Xqter)mat	U	A PROBAND	140	1	Turner's syndrome
*GM1429	8	MC5A	20U 0068	55	F	BL	47,X,+10,t(X;9)(Xpter>Xq12::9q33> 9qter;9pter>9q33::Xq12>Xqter)	U	A MOTHER	140	2	Mosaic; 16% of fibroblasts do not have additional 10
*GM0073	5	EEMEM	20U 0111		F	WH	46,X,t(X;14)(Xpter>Xq13::14q32>14qter; 14pter>14q32::Xq13>Xqter)	B	A MOTHER	147	2	
*GM0074	10	HEMEM	10U 0111	23	M	WH	47,XY,t(X;14)(Xpter>Xq13::14q32>14qter; 14pter>14q32::Xq13>Xqter)+der(14), t(X;14)(14pter>14q32::Xq13>Xqter)mat	U	A PROBAND	147	1	Klinefelter's syndrome

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME X													
*GM1730	11	EEMEM	20U	0132	5	F	BL 46,X,-21,+der(21),t(X;21)(21qter>21p11::Xq11>Xqter)mat	U	A	PROBAND	148	1	
*GM0194	15	MC5A	20U	0030	19	F	WH 46,X,t(X;3)(Xpter>Xq28::3q21>3qter;3pter>3q21::Xq28>Xqter)	B	A	PROBAND	615	1	Clinically normal; Finnish; apparently non-reciprocal trans.; structurally normal X late replicating; poor growing cul
GM2899	7	R1640	20U	0030	25	F	WH 46,X,t(X;3)(Xpter>Xq28::3q21>3qter;3pter>3q21::Xq28>Xqter)	B	A	SAME	615	1	Apparently non-reciprocal translocation; structurally normal X late replicating
*GM2859	12	HF12	20U	0036	3	F	WH 46,X,t(X;11)(Xpter>Xq11::11p11>11pter;11qter>11p11::Xq11>Xqter)	B	A	PROBAND	617	1	See GM3316 Lymphoid; South American
CHROMOSOME Y													
*GM0118	5	MC5A	20U	0052	29	F	WH 46,XX,-15,+der(15),t(Y;15)(Yqter>Yq11::15p1>15qter)pat	U	A	PROBAND			Uterine tissue fibroblast culture
GM2103	6	EEMEM	20U	0066	19	F	WH 46,X,t(X;Y)(Xpter>Xq11::Yq11>Yqter)	U	A	PROBAND			Secondary amenorrhea
GM2469	2	HF12	20U	0070	17	F	46,X,t(X;Y)(Xpter>Xq22::Yq11>Yqter)	U	A	PROBAND			
*GM2598	3	EEMEM	20U	0109	18	F	WH 46,X,t(Y;17)(Ypter>Yq11::17p13>17pter;Yqter>Yq11::17p13>17qter)	B	A	PROBAND			
CHROMOSOME 01													
*GM0097	3	MC5A	20U	0107	1	F	WH 46,X,t(X;1)(Xpter>Xq26::1q12>1qter;1pter>1q12::Xq26>Xqter)	B	A	PROBAND			De novo translocation; Beckwith-Wiedemann syndrome
*GM0126	4	MC5A	20U	0056	33	M	WH 46,XY,t(1;15)(1qter>1p36::15q1>15qter;15pter>15q1::1p36>1pter)	B	A	PROBAND			
*GM0201	10	MC5A	20U	0030	13	F	WH 47,XX,+21,t(1;17)(1pter>1p32::17p13>17qter;17pter>17p13::1p32>1qter)	U	A	PROBAND	337	1	
*GM0257	3	MC5A	20U	0002	26	F	46,XX,t(1;2)(1pter>1q32::2p23>2pter;1qter>1q32::2p23>2qter)	B	A	PROBAND			
*GM1356	2	MC5A	20U	0066	26	F	WH 46,XX,t(1;7)(1pter>1p34::7p13>7qter;7pter>7p13::1p34>1qter)	B	A	PROBAND			
*GM1421	12	MC5A	20U	0109	53	M	BL 46,XY,t(1;6)(1pter>1q42::6q21>6qter;6pter>6q21::1q42>1qter)	B	A	PROBAND			
GM1550	6	MC5A	20U	0140	31	M	WH 46,XY,t(1;3;4)(1pter>1q32::3p21>3pter;1qter>1q32::3p21>3q29::4p14>4pter;3qter>3q29::4p14>4qter)	B	A	PROBAND			

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#2	REMARKS
							TRANSLOCATION						
CHROMOSOME 01													
GM1564	9	EEMEM	20U 0151	28	F	WH	45,XX,t(1;21)(q42 or 43;q11)	U	B	PROBAND			Clinically normal
GM1813	3	HF12	20U 0066	19	F	WH	46,XX,t(1;21)(q12;q22)pat/46,XX,t(1;21)(q12;q22)pat,t(X;15)(p11;q13)	U	A	PROBAND			Menstrual dysfunction; Adj-2 segregation; Mosaic 40%/60%
GM1881	7	MC5A	20U 0140	3	M	WH	46,XY,t(1;21)(1qter>1p31::21q22>21qter; 21pter>21q22::1p31>1pter)	B	A	PROBAND			Sanfilippo A; deficient heparan N-sulfatase, normal alpha-N-acetylglucosaminidase activity
GM3501	2	HF12	20U 0140	10	M	WH	46,XY,t(1;8)(1p+;8q-)			PROBAND			Karyotype based upon lymphocytes; retardation-dysmorphic syndrome
GM1229	3	HF12	20U 0007	3	F	BL	46,XX,-1,+der(1),t(1;2)(1qter>1p36::2q31>2qter)pat	U	A	PROBAND	129	1	Multiple congenital anomalies & mental retardation
GM1230	2	HF12	20U 0007	44	M	BL	46,XY,t(1;2)(1qter>1p36::2q31>2qter; 2pter>2q31::1p36>1pter)	B	A	FATHER	129	2	Clinically normal
*GM3647	2	HF12	20U 0204	41	M	WH	46,XY,-6,+der(6),t(1;6)(6qter>6p25::1q32>1qter)mat	U	A	PROBAND	297	1	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1
*GM3648	2	HF12	20U 0204	32	M	WH	46,XY,-6,+der(6),t(1;6)(6qter>6p25::1q32>1qter)mat	U	A	HALF BRO	297	2	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1
GM3369	3	HF12	20U 0322	23	F	WH	46,XX,-1,+der(1),t(1;10)(1pter>1q44::10p11>10pter)mat	U	A	PROBAND	612	1	Developmental delay
GM3370	2	HF12	20U 0322	42	F	WH	46,XX,t(1;10)(1pter>1q44::10p11>10pter; 10qter>10p11::1q44>1qter)	B	A	MOTHER	612	2	Clinically normal
CHROMOSOME 02													
*GM0257	3	MC5A	20U 0002	26	F		46,XX,t(1;2)(1pter>1q32::2p23>2pter; 1qter>1q32::2p23>2qter)	B	A	PROBAND			
*GM0327	4	MC5A	20U 0141	24	M	WH	46,XY,t(2;8)(2pter>2q13::8q24>8qter; 8pter>8q24::2q13>2qter)	B	A	PROBAND			
GM1579	8	MC5A	20U 0030	35	F	WH	46,XX,t(2;13)(2qter>2p13::13q32>13qter;13pter>13q32::2p13>2pter)	B	A	PROBAND			Multiple malformations & mental retardation

best copy available

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 02													
GM1229	3	HF12	20U	0007	3	F	BL 46,XX,-1,+der(1),t(1;2)(1qter>1p36::2q31>2qter)pat	U	A	PROBAND	129	1	Multiple congenital anomalies & mental retardation
GM1230	2	HF12	20U	0007	44	M	BL 46,XY,t(1;2)(1qter>1p36::2q31>2qter;2pter>2q31::1p36>1pter)	B	A	FATHER	129	2	Clinically normal
*GM0501	3	MC5A	20U	0014	5	M	WH 46,XY,-2,+der(2),t(2;4)(2qter>2p25::4q21>4qter)pat	U	A	PROBAND	131	1	
*GM1064	7	MC5A	20U	0014	39	M	WH 46,XY,t(2;4)(2qter>2p25::4q21>4qter;4pter>4q21::2p25>2pter)	B	A	FATHER	131	2	
*GM0845	7	EEMEM	20U	0018	18	F	WH 46,XX,t(2;8)(2pter>2q37::8q13>8qter;8pter>8q13::2q37>2qter)mat	B	A	PROBAND	132	1	Primary amenorrhea
*GM0846	8	EEMEM	20U	0018	54	F	WH 46,XX,t(2;8)(2pter>2q37::8q13>8qter;8pter>8q13::2q37>2qter)	B	A	MOTHER	132	2	Multiple spontaneous abortions
GM1225	10	HF12	20U	0077	3MO	F	IN 46,XX,t(2;20)(2qter>2p21::20p13>20pter;20qter>20p13::2p21>2pter)	B	A	PROBAND			Indian (India); de novo; has multiple congenital anomalies
*GM0692	5	MC5A	20U	0001	8	M	WH 45,XY,-2,-21,+der(2),t(2;21)(2pter>2q37::21q2105>21qter)mat	U	A	PROBAND	133	1	Mentally retarded
*GM0693	5	MC5A	20U	0001	27	F	WH 46,XX,t(2;21)(2pter>2q37::21q2105>21qter;21pter>21q2105::2q37>2qter)	B	A	MOTHER	133	2	
GM1683	3	HF12	20U	0179	28	F	WH 46,XX,t(2;10)(p25;q26)	B	B	PROBAND	134	1	Possible red cell acid phosphatase variant
GM1848	3	HF12	20U	0179	35	M	WH 46,XY,-10,+der(10),t(2;10)(10pter>10q26::2p24>2pter)mat	U	A	MAT 1 COU	134	2	Possible red cell acid phosphatase variant
GM1849	4	HF12	20U	0179	26	M	WH 46,XY,der(10),t(2;10)(p25;q26)mat	U	B	MAT 1 COU	134	3	Possible red cell acid phosphatase variant
GM2658	4	HF12	20U	0077	31	F	WH 46,XX,t(2;6)(2pter>2q11::6q15>6qter;6pter>6q15::2q11>2qter)	B	A	PROBAND	242	1	Same patient as GM2699
CHROMOSOME 03													
GM1550	6	MC5A	20U	0140	31	M	WH 46,XY,t(1;3;4)(1pter>1q32::3p21>3pter;1qter>1q32::3p21>3q29::4p14>4pter;3qter>3q29::4p14>4qter)	B	A	PROBAND			

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 03													
*GM2808	4	HF12	20U 0140	30	F	WH	46,XX,t(3;17)(3qter>3p21::17p13>17pter; 17qter>17p13::3p21>3pter)	B	A	PROBAND			Multiple spontaneous abortions
GM2860	10	HF12	20U 0036	42	M	WH	46,XY,t(3;16)(3pter>3q132::16q13>16qter; 16pter>16q13::3q132>3qter)	B	A	PROBAND			Family history of recurrent abortions
GM2963	3	HF12	20U 0247	3	M	BL	46,XY,t(3;17)(3qter>3p21::17q25>17qter; 17pter>17q25::3p21>3pter)	B	A	PROBAND			
GM3563	2	MC5A	20U 0148	1MO	M	WH	46,XY,der(9),t(3;9)(q12;p24)mat	U	B	PROBAND			Multiple congenital anomalies
*GM0194	15	MC5A	20U 0030	19	F	WH	46,X,t(X;3)(Xpter>Xq28::3q21>3qter; 3pter>3q21::Xq28>Xqter)	B	A	PROBAND	615	1	Clinically normal; Finnish; apparently non-reciprocal trans.; structurally normal X late replicating; poor growing cul
GM2899	7	R1640	20U 0030	25	F	WH	46,X,t(X;3)(Xpter>Xq28::3q21>3qter; 3pter>3q21::Xq28>Xqter)	B	A	SAME	615	1	Apparently non-reciprocal translocation; structurally normal X late replicating
CHROMOSOME 04													
*GM0098	5	MC5A	20U 0119	10MO	M		46,XY,-(21),+der(21),t(4;21)(21qter> 21p12::4p11>4pter)mat	U	A	PROBAND			
*GM0157	12	MC5A	20U 0030	28	M	WH	49,XXXXY,t(4;11)(4pter>4q3::11q23> 11qter;11pter>11q23::4q3>4qter)	U	A	PROBAND			Severe mental retardation; sexual infantilism
*GM0380	2	MC5A	20U 0078	35	F		46,XX,t(4;11)(4pter>4q25::11q13> 11qter;11pter>11q13::4q25>4qter)	B	A	PROBAND			
*GM0624	2	MC5A	20U 0077	38	F	WH	46,XX,t(4;15)(4pter>4q11::15p11>15pter; 15qter>15p11::4q11>4qter)	B	A	PROBAND			
*GM0972	3	EEMEM	20U 0067	13	F	BL	46,XX,t(4;13)(4pter>4q31::13q14>13qter; 13pter>13q14::4q31>4qter)	B	A	PROBAND	638	1	Mental retardation; see GM1063A Lymphoid
*GM1091	3	MC5A	20U 0058	27	F	WH	46,XX,t(4;10)(4pter>4q35::10q24>10qter; 10pter>10q24::4q35>4qter)	B	A	PROBAND			
GM1101	7	EEMEM	20U 0007	32	M	WH	46,XY,t(4;12)(4qter>4p1608::12p11> 12pter;12qter>12p11::4p1608>4pter)	B	A	PROBAND			Clinically normal
GM1183	4	HF12	20U 0007	NB	M	WH	46,XY,-4,+der(4),t(4;12)(4qter> 4p1608::12p11>12pter)pat	U	A	PROBAND			
GM1550	6	MC5A	20U 0140	31	M	WH	46,XY,t(1;3;4)(1pter>1q32::3p21>3pter; 1qter>1q32::3p21>3q29::4p14>4pter; 3qter>3q29::4p14>4qter)	B	A	PROBAND			

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 04													
*GM0501	3	MC5A	20U	0014	5	M	WH 46,XY,-2,+der(2),t(2;4)(2qter>2p25::4q21>4qter)pat	U	A	PROBAND	131	1	
*GM1064	7	MC5A	20U	0014	39	M	WH 46,XY,t(2;4)(2qter>2p25::4q21>4qter;4pter>4q21::2p25>2pter)	B	A	FATHER	131	2	
*GM0773	5	EEMEM	20U	0066	26	F	46,XX,t(4;7)(4qter>4p16::7q34>7qter;7qter>7q34::4p16>4pter)	B	A	MOTHER	136	2	Clinically normal
*GM1220	3	MC5A	20U	0066	2	F	46,XX,-4,+der(4),t(4;7)(4qter>4p16::7q34>7qter)mat	U	A	PROBAND	136	1	
CHROMOSOME 05													
*GM0071	15	MC5A	20U	0015	19	F	WH 46,XX,-5,der(5),t(5;?)(p1;?)mat	U	A	PROBAND			Cri du Chat; deletion assumed to be derived rather than a simple deletion; ATCC CCL 90
*GM0344	4	MC5A	20U	0066	7	M	WH 45,XY,-5,-15,+der(5),t(5;15)(5qter>5p14::15q11>15qter),t(9;11)(9qter>9p24::11q23>11qter;11pter>11q23::9p24>9pter)	U	A	PROBAND			Cri du Chat
*GM0589	2	MC5A	20U	0012	18	M	WH 46,XY,t(5;14)(5qter>5p14::14q21>14qter;14pter>14q21::5p14>5pter)	B	A	PROBAND			
GM1678	3	HF12	20U	0140	33	F	46,XX,t(5;10)(5qter>5p15::10p13>10pter;10qter>10p13::5p15>5pter)	B	A	PROBAND			
*GM2426	5	EEMEM	20U	0134	12	M	WH 46,XY,t(5;6)(5pter>5p15::6p21>6qter;6pter>6p21::5p15>5qter)	B	A	PROBAND			Clinically normal
GM3493	2	HF12	20U	0204	5MO	F	WH 46,XX,-5,+der(5),t(5;6)(5pter>5q35::6p22>6pter)pat	U	A	PROBAND	135	1	Partial trisomy 6p; psychomotor retardation; multiple congenital anomalies
GM3494A	5	EEMEM	20U	0204	27	M	WH 46,XY,t(5;6)(5pter>5q35::6p22>6pter;5qter>5q35::6p22>6qter)	B	A	FATHER	135	2	Clinically normal; balanced carrier
GM1535	6	MC5A	20U	0002	16	F	BL 46,XX,-12,+der(12),t(5;12)(12pter>12q24::5q33>5qter)mat	U	A	PROBAND	139	1	
GM1536	6	EEMEM	20U	0002	38	F	BL 46,XX,t(5;12)(5pter>5q33::12q24>12qter;12pter>12q24::5q33>5qter)	B	A	MOTHER	139	2	
GM2487		MC5A	20U	0140	44	F	WH 46,XX,t(5;15)(5pter>5p13::15p11>15qter;15pter>15p11::5p13>5qter)	B	A	MOTHER	225	2	Passage 5 at IMR; translocation is present in all cells, different cells show different aberrations including X chromosome aberrations

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
TRANSLOCATION												
CHROMOSOME 05												
GM2488		MC5A	20U 0140	23	M	WH	46,XY,-15,+der(15),t(5;15)(5pter>5p13::15p11>15qter)mat	U	A PROBAND	225	1	Passage 6 at IMR
*GM3305	4	HF12	20U 0204	28	M	WH	46,XY,t(5;9)(5pter>5q33::9p24>9pter;9qter>9p24::5q33>5qter)	B	A FATHER	611	2	
CHROMOSOME 06												
*GM0144	5	MC5A	20U 0012	34	F	WH	46,XX,-6,+der(6),t(6;21)(6qter>6p25::21q11>21qter)mat	U	A PROBAND			13% of cells found to be balanced in passage 1 after recovery; these tend to increase with passage in culture
*GM0610	7	MC5A	20U 0075	11WK	F	WH	46,XX,t(6;18)(6pter>6q21::18p11>18pter;6qter>6q21::18p11>18qter)	B	A PROBAND			
*GM1137	6	R1640	10U 0017	1MO	M	BL	48,XXY,+21,t(6;10)(6qter>6p22 or 24::10p12>10pter;10qter>10p12::6p22 or 24>6pter)	U	B PROBAND			Down's syndrome features
*GM1139	4	MC5A	20U 0095	21	F	WH	46,XX,t(6;13)(6qter>6p21::13q34>13qter;13pter>13q34::6p21>6pter),t(15;17)(q15;p13)	B	A PROBAND			Culture is mosaic: 64% carry only the t(15;17), 36% carry both
*GM1421	12	MC5A	20U 0109	53	M	BL	46,XY,t(1;6)(1pter>1q42::6q21>6qter;6pter>6q21::1q42>1qter)	B	A PROBAND			
*GM1605	3	MC5A	20U 0166	34	M	WH	46,XY,t(6;11)(6qter>6p23::11q23>11qter;11pter>11q23::6p23>6pter)	B	A PROBAND			
GM2068	5	EEMEM	10U 0088	4	F	WH	46,XX,t(6;7)(6pter>6q27::7q22>7qter;7pter>7q22::6q27>6qter)	B	A PROBAND			
*GM2426	5	EEMEM	20U 0134	12	M	WH	46,XY,t(5;6)(5pter>5p15::6p21>6qter;6pter>6p21::5p15>5qter)	B	A PROBAND			Clinically normal
GM2594	4	EEMEM	20U 0073	30	M	WH	46,XY,t(6;9)(6pter>6q13::9p22>9pter;9qter>9p22::6q13>6qter)	B	A PROBAND			Habitual aborter
GM3404	2	HF12	20U 0066	28	F		46,XX,t(6;12)(6pter>6q21::12p13>12pter;6qter>6q21::12p13>12qter)	B	A PROBAND			
GM3493	2	HF12	20U 0204	5MO	F	WH	46,XX,-5,+der(5),t(5;6)(5pter>5q35::6p22>6pter)pat	U	A PROBAND	135	1	Partial trisomy 6p; psychomotor retardation; multiple congenital anomalies
GM3494A	5	EEMEM	20U 0204	27	M	WH	46,XY,t(5;6)(5pter>5q35::6p22>6pter;5qter>5q35::6p22>6qter)	B	A FATHER	135	2	Clinically normal; balanced carrier

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 06													
GM2658	4	HF12	20U 0077	31	F	WH	46,XX,t(2;6)(2pter>2q11::6q15>6qter; 6pter>6q15::2q11>2qter)	B	A	PROBAND	242	1	Same patient as GM2699
*GM3647	2	HF12	20U 0204	41	M	WH	46,XY,-6,+der(6),t(1;6)(6qter>6p25::1q32>1qter)mat	U	A	PROBAND	297	1	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1
*GM3648	2	HF12	20U 0204	32	M	WH	46,XY,-6,+der(6),t(1;6)(6qter>6p25::1q32>1qter)mat	U	A	HALF BRO	297	2	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1
CHROMOSOME 07													
*GM0044	5	MC5A	20U 0097	6	M	WH	46,XY,t(7;10)(7qter>7p2::10q11>10qter; 10pter>10q11::7p2>7pter)	B	A	PROBAND			Congenital malformations, severe retardation although apparently balanced karyotype
*GM0657	3	MC5A	20U 0107	4WK	M	WH	46,XY,t(7;18)(7pter>7q36::18q21>18qter; 18pter>18q21::7q36>7qter)	B	A	PROBAND			
*GM1356	2	MC5A	20U 0066	26	F	WH	46,XX,t(1;7)(1pter>1p34::7p13>7qter; 7pter>7p13::1p34>1qter)	B	A	PROBAND			
*GM1696	9	EEMEM	20U 0176	30	F	WH	46,X,t(X;7)(Xpter>Xq21::7p22>7pter; Xqter>Xq21::7p22>7qter)	B	A	PROBAND			
GM2068	5	EEMEM	10U 0088	4	F	WH	46,XX,t(6;7)(6pter>6q27::7q22>7qter; 7pter>7q22::6q27>6qter)	B	A	PROBAND			
*GM0773	5	EEMEM	20U 0066	26	F		46,XX,t(4;7)(4qter>4p16::7q34>7qter; 7qter>7q34::4p16>4pter)	B	A	MOTHER	136	2	Clinically normal
*GM1220	3	MC5A	20U 0066	2	F		46,XX,-4,+der(4),t(4;7)(4qter>4p16::7q34>7qter)mat	U	A	PROBAND	136	1	
CHROMOSOME 08													
*GM0213	5	MC5A	20U 0082	35	M	WH	46,XY,t(8;12)(8qter>8p23::12p11>12pter; 12qter>12p11::8p23>8pter)	B	A	PROBAND			
*GM0327	4	MC5A	20U 0141	24	M	WH	46,XY,t(2;8)(2pter>2q13::8q24>8qter; 8pter>8q24::2q13>2qter)	B	A	PROBAND			
GM1512	6	MC5A	20U 0030	26	M	WH	46,XY,t(8;10)(8pter>8p21::10p13>10qter; 10pter>10p13::8p21>8pter)	B	B	PROBAND			
GM3501	2	HF12	20U 0140	10	M	WH	46,XY,t(1;8)(1p+;8q-)			PROBAND			Karyotype based upon lymphocytes; retardation-dysmorphic syndrome

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
TRANSLOCATION													
CHROMOSOME 08													
*GM3689	6	EEMEM	20U 0247	24	F	WH	46,XX,-10,+der(10),t(8;10)(8pter>8p21.1::10p15>10qter)mat	U	A	PROBAND			Mental & growth retardation; delayed sexual maturation; multiple congenital anomalies
*GM0845	7	EEMEM	20U 0018	18	F	WH	46,XX,t(2;8)(2pter>2q37::8q13>8qter;8pter>8q13::2q37>2qter)mat	B	A	PROBAND	132	1	Primary amenorrhea
*GM0846	8	EEMEM	20U 0018	54	F	WH	46,XX,t(2;8)(2pter>2q37::8q13>8qter;8pter>8q13::2q37>2qter)	B	A	MOTHER	132	2	Multiple spontaneous abortions
GM3256	3	HF12	20U 0204	25	F	WH	46,XX,t(8;15)(8qter>8p21::15q26>15qter;15pter>15q26::8p21>8pter)	B	A	MOTHER	609	2	
GM3255	3	HF12	20U 0204	5	M	WH	46,XY,-15,+der(15),t(8;15)(15pter>15q26::8p21>8pter)mat	U	A	PROBAND	609	1	Partial trisomy 8(p);
CHROMOSOME 09													
*GM0344	4	MC5A	20U 0066	7	M	WH	45,XY,-5,-15,+der(5),t(5;15)(5qter>5p14::15q11>15qter),t(9;11)(9qter>9p24::11q23>11qter;11pter>11q23::9p24>9pter)	U	A	PROBAND			Cri du Chat
*GM0705	13	HF12	20U 0016	10MO	F	WH	46,X,t(X;9)(Xpter>Xq12::9p24>9pter;9qter>9p24::Xq12>Xqter)	B	A	PROBAND			Culture grows slowly
*GM0988	10	HF12	10U 0082	3WK	M	WH	46,XY,t(9;17)(9qter>9p13::17q25>17qter;17pter>17q25::9p13>9pter)	B	A	PROBAND			Multiple congenital anomalies; 13% of cells are polyploid
*GM1387	4	MC5A	20U 0061	14	F	WH	46,XX,-13,+der(9),t(9;13)(q22;q12)mat	U	B	PROBAND	369	1	Result of Adj-2 segregation; see also GM1388 Lymphoid
*GM1664	6	EEMEM	20U 0113	40	M	WH	46,XY,t(9;13)(9pter>9q13::13q12>13qter;13pter>13q12::9q13>9qter)	B	A	PROBAND			
GM1734	3	HF12	20U 0140	40	M		46,XY,der(15),t(9;15)(p11;q11)mat	U	B	PROBAND	257	1	
GM1892	5	HF12	20U 0132	37	F	WH	46,XX,t(9;18)(9pter>9p24::18q12>18qter;18pter>18q12::9p24>9pter)	B	A	PROBAND			
*GM2492	6	HF12	20U 0221	38	M	WH	46,XY,t(9;13)(9qter>9p22 or p23::13q14 or q21>13qter;13pter>13q14 or q21::9p22 or p23>9pter)	B	A	PROBAND			
GM2594	4	EEMEM	20U 0073	30	M	WH	46,XY,t(6;9)(6pter>6q13::9p22>9pter;9qter>9p22::6q13>6qter)	B	A	PROBAND			Habitual aborter

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 09													
GM2836	4	HF12	20U 0263	43	M	WH	46,XY,t(9;17)(q12;p11)/46,XY,t(9;17)(q12;p11),ins(3;1)(q21;q25q44)	B	A	PROBAND			61%/29%
GM3268	2	HF12	20U 0122	10MO	M	BL	46,XY,-21,+der(21),t(9;21)	U		PROBAND			Karyotype based upon lymphocytes
GM3563	2	MC5A	20U 0148	1MO	M	WH	46,XY,der(9),t(3;9)(q12;p24)mat	U	B	PROBAND			Multiple congenital anomalies
*GM1414	5	EEMEM	20U 0068	28	F	BL	46,X,+der(9),t(X;9)(9pter>9q34::Xq13>Xqter)mat	U	A	PROBAND	140	1	Turner's syndrome
*GM1429	8	MC5A	20U 0068	55	F	BL	47,X,+10,t(X;9)(Xpter>Xq12::9q33>9qter;9pter>9q33::Xq12>Xqter)	U	A	MOTHER	140	2	Mosaic; 16% of fibroblasts do not have additional 10
GM1750	2	HF12	20U 0070	30MO	M	WH	47,XY,+der(14),t(9;14)(p24;q22)mat	U	B	PROBAND	141	1	
GM1751	2	HF12	20U 0070	26	F	WH	46,XX,t(9;14)(p24;q22)	B	B	MOTHER	141	2	
GM3462	2	HF12	20U 0204	3MO	F	WH	47,XX,+der(14),t(9;14)(14pter>14q13::9p22>9pter)mat	U	A	PROBAND	227	1	Growth retardation and congenital malformations
GM3463	4	HF12	20U 0204	23	F	WH	46,XX,t(9;14)(9qter>9p22::14q13>14qter)(14pter>14q13::9p22>9pter)	B	A	MOTHER	227	2	Clinically normal
GM2819	3	HF12	20U 0107	7MO	F	BL	46,XX,-12,+der(12),t(9;12)(12pter>12q24::9p13>9pter)mat	U	A	PROBAND	324	1	See GM2820 Lymphoid
GM2821	2	HF12	20U 0107		F	BL	46,XX,t(9;12)(9qter>9p13::12q24>12qter;12pter>12q24::9p13>9pter)	B	A	MOTHER	324	2	See GM2822 Lymphoid
GM3119	4	MC5A	20U 0172	28	F	WH	46,XX,t(9;17)(9pter>9p22::17p11>17qter;17pter>17p11::9p22>9qter)	B	A	MOTHER	449	2	Fetus is GM3100 Amniotic
*GM3305	4	HF12	20U 0204	28	M	WH	46,XY,t(5;9)(5pter>5q33::9p24>9pter;9qter>9p24::5q33>5qter)	B	A	FATHER	611	2	
CHROMOSOME 10													
*GM0044	5	MC5A	20U 0097	6	M	WH	46,XY,t(7;10)(7qter>7p2::10q11>10qter;10pter>10q11::7p2>7pter)	B	A	PROBAND			Congenital malformations, severe retardation although apparently balanced karyotype
*GM1091	3	MC5A	20U 0058	27	F	WH	46,XX,t(4;10)(4pter>4q35::10q24>10qter;10pter>10q24::4q35>4qter)	B	A	PROBAND			

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION					
CHROMOSOME 10												
*GM1137	6	R1640	10U	0017	1MO	M	BL 48,XXY,+21,t(6;10)(6qter>6p22 or 24::10p12>10pter;10qter>10p12::6p22 or 24>6pter)	U	B PROBAND			Down's syndrome features
*GM1396	3	MC5A	20U	0095	3	M	WH 46,XY,-10,+der(10),t(10;16)(10pter>10q26::16q22>16qter)pat	U	A PROBAND			
GM1512	6	MC5A	20U	0030	26	M	WH 46,XY,t(8;10)(8pter>8p21::10p13>10qter;10pter>10p13::8p21>8pter)	B	B PROBAND			
GM1678	3	HF12	20U	0140	33	F	46,XX,t(5;10)(5qter>5p15::10p13>10pter;10qter>10p13::5p15>5pter)	B	A PROBAND			
GM3241A	3	HF12	20U	0016	10MO	F	WH 45,XX,t(10;13)(q26;q13)	U	PROBAND			Parents both have normal karyotypes; karyotype based upon lymphocytes
*GM3689	6	EEMEM	20U	0247	24	F	WH 46,XX,-10,+der(10),t(8;10)(8pter>8p21.1::10p15>10qter)mat	U	A PROBAND			Mental & growth retardation; delayed sexual maturation; multiple congenital anomalies
GM1683	3	HF12	20U	0179	28	F	WH 46,XX,t(2;10)(p25;q26)	B	B PROBAND	134	1	Possible red cell acid phosphatase variant
GM1848	3	HF12	20U	0179	35	M	WH 46,XY,-10,+der(10),t(2;10)(10pter>10q26::2p24>2pter)mat	U	A MAT 1 COU	134	2	Possible red cell acid phosphatase variant
GM1849	4	HF12	20U	0179	26	M	WH 46,XY,der(10),t(2;10)(p25;q26)mat	U	B MAT 1 COU	134	3	Possible red cell acid phosphatase variant
*GM0216	4	MC5A	20U	0143	29	M	WH 46,XY,t(10;17)(10pter>10q24::17p13>17pter;17qter>17p13::10q24>10qter)	B	A FATHER	142	2	
*GM0217	6	MC5A	20U	0143	9DA	M	WH 46,XY,-17,+der(17),t(10;17)(17qter>17p13::10q24>10qter)pat	U	A PROBAND	142	1	Culture grows slowly
*GM0959	6	EEMEM	20U	0143	11	M	WH 46,XY,-17,+der(17),t(10;17)(17qter>17p13::10q24>10qter)pat	U	A MAT 3RD COU	142	3	
*GM1399	5	HF12	20U	0132	20	M	BL 47,XY,+der(21),t(10;21)(21pter>21q2105::10q26>10qter)mat	U	A MAT 1 COU	143	3	Mental retardation and ear anomalies; 1st cousin once removed
*GM1413	8	HF12	20U	0132	18MO	F	BL 46,XX,-10,+der(10),t(10;21)(10pter>10q26::21q2105>21qter)mat	U	A PROBAND	143	1	Down's syndrome

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 10													
*GM1580	6	HF12	20U	0132	48	F	BL 46,XX,t(10;21)(10pter>10q26::21q21>21qter)(21pter>21q21::10q26>10qter)	B	A	MAT GRMR	143	2	Normal phenotype
*GM0983	3	EEMEM	20U	0095	14MO	M	WH 46,XY,-21,+der(21),t(10;21)(10qter>10q24::21q22>21pter)mat	U	A	PROBAND	144	1	
*GM0984	3	EEMEM	20U	0095	34	F	WH 46,XX,t(10;21)(10pter>10q24::21q22>21qter;21pter>21q22::10q24>10qter)	B	A	MOTHER	144	2	
GM3369	3	HF12	20U	0322	23	F	WH 46,XX,-1,+der(1),t(1;10)(1pter>1q44::10p11>10pter)mat	U	A	PROBAND	612	1	Developmental delay
GM3370	2	HF12	20U	0322	42	F	WH 46,XX,t(1;10)(1pter>1q44::10p11>10pter;10qter>10p11::1q44>1qter)	B	A	MOTHER	612	2	Clinically normal
CHROMOSOME 11													
*GM0157	12	MC5A	20U	0030	28	M	WH 49,XXXXY,t(4;11)(4pter>4q3::11q23>11qter;11pter>11q23::4q3>4qter)	U	A	PROBAND			Severe mental retardation; sexual infantilism
*GM0344	4	MC5A	20U	0066	7	M	WH 45,XY,-5,-15,+der(5),t(5;15)(5qter>5p14::15q11>15qter),t(9;11)(9qter>9p24::11q23>11qter;11pter>11q23::9p24>9pter)	U	A	PROBAND			Cri du Chat
*GM0380	2	MC5A	20U	0078	35	F	46,XX,t(4;11)(4pter>4q25::11q13>11qter;11pter>11q13::4q25>4qter)	B	A	PROBAND			
*GM0980	2	MC5A	20U	0012	33	F	WH 45,XX,-11,-22,+der(11),t(11;22)(11pter>11q25::22q11>22qter)	U	A	PROBAND			69% of cells also show t(1;4)(q25;p12)
*GM1605	3	MC5A	20U	0166	34	M	WH 46,XY,t(6;11)(6qter>6p23::11q23>11qter;11pter>11q23::6p23>6pter)	B	A	PROBAND			
*GM1695	5	HF12	20U	0176	26	F	WH 46,X,t(X;11)(Xqter>Xp2105::11q13>11qter;11pter>11q13::Xp2105>Xpter)	B	A	PROBAND			Also has clinically documented Duchenne Muscular Dystrophy
GM3005	8	EEMEM	20U	0268	28	F	WH 46,XX,t(11;16)(11pter>11q13::16p11>16pter;11qter>11q13::16p11>16qter)	B	A	PROBAND			
*GM3322	4	HF12	20U	0070	35	F	WH 46,X,t(X;11)(Xpter>Xq22::11q13>11qter;11pter>11q13::Xq22>Xqter)	B	A	PROBAND			
GM3552	7	EEMEM	20U	0096	29	F	OR 46,X,t(X;11)(Xpter>Xq26::11q23>11qter;11pter>11q23::Xq26>Xqter)	B	A	PROBAND			Vietnamese; gonadal dysgenesis
GM3371	2	HF12	20U	0322	SWK	F	BL 47,XX,t(11;22)(q25;q13),+der(22),t(11;22)(22pter>22q13::11q25>11qter)mat,inv(9)(p13;q12)pat	U	A	PROBAND	613	1	Partial trisomy 22; father has same inv(9); developmental delay

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
TRANSLOCATION													
CHROMOSOME 11													
GM3372	2	HF12	20U 0322	28	F	BL	46,XX,t(11;22)(11pter>11q25::22q13>22qter;22pter>22q13::11q25>11qter)	B	A	MOTHER	613	2	Clinically normal
*GM2859	12	HF12	20U 0036	3	F	WH	46,X,t(X;11)(Xpter>Xq11::11p11>11pter;11qter>11p11::Xq11>Xqter)	B	A	PROBAND	617	1	See GM3316 Lymphoid; South American
CHROMOSOME 12													
*GM0213	5	MC5A	20U 0082	35	M	WH	46,XY,t(8;12)(8qter>8p23::12p11>12pter;12qter>12p11::8p23>8pter)	B	A	PROBAND			
GM1101	7	EEMEM	20U 0007	32	M	WH	46,XY,t(4;12)(4qter>4p1608::12p11>12pter;12qter>12p11::4p1608>4pter)	B	A	PROBAND			Clinically normal
GM1183	4	HF12	20U 0007	NB	M	WH	46,XY,-4,+der(4),t(4;12)(4qter>4p1608::12p11>12pter)pat	U	A	PROBAND			
GM1665	3	HF12	20U 0140	28	F	WH	46,XX,t(12;21)(12qter>12p11::21p11>21pter;21qter>21p11::12p11>12pter)	B	A	PROBAND			
*GM2621	8	EEMEM	10U 0224	32	F	WH	46,X,t(X;12)(Xpter>Xq22::12q24>12qter;12pter>12q24::Xq22>Xqter)	B	A	PROBAND			Gonadal dysgenesis
GM3404	2	HF12	20U 0066	28	F		46,XX,t(6;12)(6pter>6q21::12p13>12pter;6qter>6q21::12p13>12qter)	B	A	PROBAND			
GM1535	6	MC5A	20U 0002	16	F	BL	46,XX,-12,+der(12),t(5;12)(12pter>12q24::5q33>5qter)mat	U	A	PROBAND	139	1	
GM1536	6	EEMEM	20U 0002	38	F	BL	46,XX,t(5;12)(5pter>5q33::12q24>12qter;12pter>12q24::5q33>5qter)	B	A	MOTHER	139	2	
GM2819	3	HF12	20U 0107	7MO	F	BL	46,XX,-12,+der(12),t(9;12)(12pter>12q24::9p13>9pter)mat	U	A	PROBAND	324	1	See GM2820 Lymphoid
GM2821	2	HF12	20U 0107		F	BL	46,XX,t(9;12)(9qter>9p13::12q24>12qter;12pter>12q24::9p13>9pter)	B	A	MOTHER	324	2	See GM2822 Lymphoid
CHROMOSOME 13													
*GM0085	6	MC5A	20U 0143	2WK	M	BL	46,XY,-13,+t(13;13)(13qter>cen>13qter)	U	A	PROBAND			Clinical Trisomy 13
*GM0392	9	MC5A	20U 0002	1	F	WH	45,XX,t(13;22)(13qter>cen>22qter)	U	A	PROBAND			
*GM0627	3	MC5A	20U 0058	30	F	BL	46,XX,t(13;22)(13pter>13q22::22q13>22qter;22pter>22q13::13q22>13qter)	B	A	PROBAND			
*GM0972	3	EEMEM	20U 0067	13	F	BL	46,XX,t(4;13)(4pter>4q31::13q14>13qter;13pter>13q14::4q31>4qter)	B	A	PROBAND	638	1	Mental retardation; see GM1063A Lymphoid

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 13													
*GM1139	4	MC5A	20U 0095	21	F	WH	46,XX,t(6;13)(6qter>6p21::13q34>13qter; 13pter>13q34::6p21>6pter),t(15;17)(q15; p13)	B	A	PROBAND			Culture is mosaic: 64% carry only the t(15;17), 36% carry both
*GM1224	7	HF12	20U 0077	3MO	F	WH	46,XX,-13,+der(13),t(13; 8)(13pter> 13q34::18q11>18qter)pat	U	A	PROBAND			
GM1296	4	EEMEM	20U 0012	47	F	WH	45,XX,t(13;15)(13qter>cen>15qter)	U	A	PROBAND			Clinically normal; in addition to t(13;15) some cells show 21p+ and 10q- which could be a t(10;21)(q24;p12)
*GM1387	4	MC5A	20U 0061	14	F	WH	46,XX,-13,+der(9),t(9;13)(q22;q12)mat	U	B	PROBAND	369	1	Result of Adj-2 segregation; see also GM1388 Lymphoid
GM1579	8	MC5A	20U 0030	35	F	WH	46,XX,t(2;13)(2qter>2p13::13q32> 13qter;13pter>13q32::2p13>2pter)	B	A	PROBAND			Multiple malformations & mental retardation
*GM1664	6	EEMEM	20U 0113	40	M	WH	46,XY,t(9;13)(9pter>9q13::13q12>13qter; 13pter>13q12::9q13>9qter)	B	A	PROBAND			
*GM2018	12	HF12	20U 0077	2	F	WH	45,XX,tan(13;13)(pter>q34::q12>qter)	U	A	PROBAND			
*GM2492	6	HF12	20U 0221	38	M	WH	46,XY,t(9;13)(9qter>9p22 or p23::13q14 or q21>13qter;13pter>13q14 or q21::9p22 or p23>9pter)	B	A	PROBAND			
GM2813	7	R1640	20U 0148	1WK	F	WH	46,XX,-15,+der(13q15q),t(13;15)(13qter> cen>15qter)mat	U	A	PROBAND			
GM3089	6	MC5A	20U 0299	3WK	F	WH	46,XX,-18,+der(18),t(13;18)(13qter> 13q31::18q23>18pter)pat	U	A	PROBAND	301	1	
GM3241A	3	HF12	20U 0016	10MO	F	WH	45,XX,t(10;13)(q26;q13)	U		PROBAND			Parents both have normal karyotypes; karyotype based upon lymphocytes
*GM1555	5	EEMEM	20U 0113	13	M	WH	47,XY,+der(13),t(13;17)(13pter>13q14:: 17p13>17pter)mat	U	A	BRO	145	2	
*GM1663	13	MC5A	20U 0113	14	M	WH	46,XY,-17,+der(17),t(13;17)(17qter> 17p13::13q14>13qter)mat	U	A	PROBAND	145	1	Partial trisomy 13
CHROMOSOME 14													
*GM0005	5	MC5A	20U 0111	30	F	WH	45,XX,t(14;22)(14qter>cen>22qter)	U	A	PROBAND			

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 14													
*GM0479	12	EEMEM	20U 0048	35	F	WH	45,XX,t(14;15)(14qter>cen>15qter)	U	A	PROBAND			
*GM0589	2	MC5A	20U 0012	18	M	WH	46,XY,t(5;14)(5qter>5p14::14q21>14qter; 14pter>14q21::5p14>5pter)	B	A	PROBAND			
GM2044	2	HF12	20U 0164	7	M		45,XY,-16,t(14;16)(16;18)(14qter>14p12:: 16p11>16pter;18qter>18p1::16q12>16qter)	U	A	PROBAND			
GM1750	2	HF12	20U 0070	30MO	M	WH	47,XY,+der(14),t(9;14)(p24;q22)mat	U	B	PROBAND	141	1	
GM1751	2	HF12	20U 0070	26	F	WH	46,XX,t(9;14)(p24;q22)	B	B	MOTHER	141	2	
*GM0981	3	EEMEM	20U 0148	4	F	BL	46,XX,-14,+der(14),t(14;20)(14qter> 14p11::20p11>20pter)mat,inv(9)(pter> p13::q13>p13::q13>qter)	U	A	PROBAND	146	1	
*GM0982	3	EEMEM	20U 0148		F	BL	46,XX,t(14;20)(14pter>14p11::20p11> 20qter;20pter>20p11::14p11>14qter)	B	A	MOTHER	146	2	
*GM0073	5	EEMEM	20U 0111		F	WH	46,X,t(X;14)(Xpter>Xq13::14q32>14qter; 14pter>14q32::Xq13>Xqter)	B	A	MOTHER	147	2	
*GM0074	10	HEMEM	10U 0111	23	M	WH	47,XY,t(X;14)(Xpter>Xq13::14q32>14qter; 14pter>14q32::Xq13>Xqter)+der(14), t(X;14)(14pter>14q32::Xq13>Xqter)mat	U	A	PROBAND	147	1	Klinefelter's syndrome
GM3462	2	HF12	20U 0204	3MO	F	WH	47,XX,+der(14),t(9;14)(14pter>14q13:: 9p22>9pter)mat	U	A	PROBAND	227	1	Growth retardation and congenital malformations
GM3463	4	HF12	20U 0204	23	F	WH	46,XX,t(9;14)(9qter>9p22::14q13>14qter) (14pter>14q13::9p22>9pter)	B	A	MOTHER	227	2	Clinically normal
CHROMOSOME 15													
*GM0017	4	MC5A	20U 0012	30MO	M	BL	45,XY,-15,-18,+der(18),t(15;18)(18pter> 18q23::15q1>15qter)	U	A	PROBAND			Multiple congenital anomalies & mental retardation
*GM0118	5	MC5A	20U 0052	29	F	WH	46,XX,-15,+der(15),t(Y;15)(Yqter>Yq11:: 15p1>15qter)pat	U	A	PROBAND			Uterine tissue fibroblast culture
*GM0126	4	MC5A	20U 0056	33	M	WH	46,XY,t(1;15)(1qter>1p36::15q1>15qter; 15pter>15q1::1p36>1pter)	B	A	PROBAND			
*GM0344	4	MC5A	20U 0066	7	M	WH	45,XY,-5,-15,+der(5),t(5;15)(5qter>5p14 ::15q11>15qter),t(9;11)(9qter>9p24:: 11q23>11qter;11pter>11q23::9p24>9pter)	U	A	PROBAND			Cri du Chat

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 15													
*GM0479	12	EEMEM	20U	0048	35	F	WH 45,XX,t(14;15)(14qter>cen>15qter)	U	A	PROBAND			
*GM0624	2	MC5A	20U	0077	38	F	WH 46,XX,t(4;15)(4pter>4q11::15p11>15pter; 15qter>15p11::4q11>4qter)	B	A	PROBAND			
*GM1139	4	MC5A	20U	0095	21	F	WH 46,XX,t(15;17)(15pter>15q15::17p13> 17pter;17qter>17p13::15q15>15qter), t(6;13)(p21;q34)	B	A	PROBAND			Culture is mosaic: 64% carry only the t(15;17), 36% carry both
GM1296	4	EEMEM	20U	0012	47	F	WH 45,XX,t(13;15)(13qter>cen>15qter)	U	A	PROBAND			Clinically normal; in addition to t(13;15) some cells show 21p+ and 10q- which could be a t(10;21) (q24;p12)
GM1734	3	HF12	20U	0140	40	M	46,XY,der(15),t(9;15)(p11;q11)mat	U	B	PROBAND	257	1	
GM1813	3	HF12	20U	0066	19	F	WH 46,XX,t(1;21)(q12;q22)pat/46,XX,t(1;21) (q12;q22)pat,t(X;15)(p11;q13)	U	A	PROBAND			Menstrual dysfunction; Adj-2 segregation; Mosaic 40%/60%
GM2729	2	MC5A	20U	0116	15	M	WH 47,XY,+t(15;15)(15pter>15q1?::15q1?> ?cen>15pter)	U	B	PROBAND			
GM2813	7	R1640	20U	0148	1WK	F	WH 46,XX,-15,+der(13q15q),t(13;15)(13qter> cen>15qter)mat	U	A	PROBAND			
GM2487		MC5A	20U	0140	44	F	WH 46,XX,t(5;15)(5pter>5p13::15p11>15qter; 15pter>15p11::5p13>5qter)	B	A	MOTHER	225	2	Passage 5 at IMR; translocation is present in all cells, different cells show different aberrations including X chromosome aberrations
GM2488		MC5A	20U	0140	23	M	WH 46,XY,-15,+der(15),t(5;15)(5pter>5p13:: 15p11>15qter)mat	U	A	PROBAND	225	1	Passage 6 at IMR
GM3255	3	HF12	20U	0204	5	M	WH 46,XY,-15,+der(15),t(8;15)(15pter> 15q26::8p21>8pter)mat	U	A	PROBAND	609	1	Partial trisomy 8(p);
GM3256	3	HF12	20U	0204	25	F	WH 46,XX,t(8;15)(8qter>8p21::15q26:15qter; 15pter>15q26::8p21>8pter)	B	A	MOTHER	609	2	
CHROMOSOME 16													
*GM1396	3	MC5A	20U	0095	3	M	WH 46,XY,-10,+der(10),t(10;16)(10pter> 10q26::16q22>16qter)pat	U	A	PROBAND			
GM2044	2	HF12	20U	0164	7	M	45,XY,-16,t(14;16)(16;18)(14qter>14p12:: 16p11>16pter;18qter>18p1::16q12>16qter)	U	A	PROBAND			

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION					
CHROMOSOME 16												
GM2860	10	HF12	20U 0036	42	M	WH	46,XY,t(3;16)(3pter>3q132::16q13>16qter; 16pter>16q13::3q132>3qter)	B	A PROBAND			Family history of recurrent abortions
GM3005	8	EEMEM	20U 0268	28	F	WH	46,XX,t(11;16)(11pter>11q13::16p11> 16pter;11qter>11q13::16p11>16qter)	B	A PROBAND			
GM2325	4	HF12	20U 0182	11DA	F	WH	47,XX,+der(22),t(16;22)(22pter>22q12:: 16p13>16pter)mat	U	A PROBAND	179	1	Daughter of GM2324 Lymphoid
CHROMOSOME 17												
*GM0119	5	MC5A	20U 0087	31	F	WH	46,XX,t(17;22)(17qter>17p13::22q11> 22qter;22pter>22q11::17p13>17pter)	B	A PROBAND			Clinically normal
*GM0201	10	MC5A	20U 0030	13	F	WH	47,XX,+21,t(1;17)(1pter>1p32::17p13> 17qter;17pter>17p13::1p32>1qter)	U	A PROBAND	337	1	
*GM0271	3	MC5A	20U 0058	28	F	WH	46,XX,t(17;19)(17pter>17q23::19p13> 19pter;19qter>19p13::17q23>17qter)	B	A PROBAND			Normal phenotype; 1 spontaneous abortion and 1 stillborn with anomalies
*GM0988	10	HF12	10U 0082	3WK	M	WH	46,XY,t(9;17)(9qter>9p13::17q25>17qter; 17pter>17q25::9p13>9pter)	B	A PROBAND			Multiple congenital anomalies; 13% of cells are polyploid
*GM1139	4	MC5A	20U 0095	21	F	WH	46,XX,t(15;17)(15pter>15q15::17p13> 17pter;17qter>17p13::15q15>15qter), t(6;13)(p21;q34)	B	A PROBAND			Culture is mosaic: 64% carry only the t(15;17), 36% carry both
*GM2598	3	EEMEM	20U 0109	18	F	WH	46,X,t(Y;17)(Ypter>Yq11::17p13>17pter; Yqter>Yq11::17p13>17qter)	B	A PROBAND			
*GM2808	4	HF12	20U 0140	30	F	WH	46,XX,t(3;17)(3qter>3p21::17p13>17pter; 17qter>17p13::3p21>3pter)	B	A PROBAND			Multiple spontaneous abortions
GM2836	4	HF12	20U 0263	43	M	WH	46,XY,t(9;17)(q12;p11)/46,XY,t(9;17) (q12;p11),ins(3;1)(q21;q25q44)	B	A PROBAND			61%/29%
GM2963	3	HF12	20U 0247	3	M	BL	46,XY,t(3;17)(3qter>3p21::17q25>17qter; 17pter>17q25::3p21>3pter)	B	A PROBAND			
*GM0216	4	MC5A	20U 0143	29	M	WH	46,XY,t(10;17)(10pter>10q24::17p13> 17pter;17qter>17p13::10q24>10qter)	B	A FATHER	142	2	
*GM0217	6	MC5A	20U 0143	9DA	M	WH	46,XY,-17,+der(17),t(10;17)(17qter> 17p13::10q24>10qter)pat	U	A PROBAND	142	1	Culture grows slowly
*GM0959	6	EEMEM	20U 0143	11	M	WH	46,XY,-17,+der(17),t(10;17)(17qter> 17p13::10q24>10qter)pat	U	A MAT 3RD COU	142	3	

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
TRANSLOCATION													
CHROMOSOME 17													
*GM1555	5	EEMEM	20U 0113	13	M	WH	47,XY,+der(13),t(13;17)(13pter>13q14::17p13>17pter)mat	U	A	BRO	145	2	
*GM1663	13	MC5A	20U 0113	14	M	WH	46,XY,-17,+der(17),t(13;17)(17qter>17p13::13q14>13qter)mat	U	A	PROBAND	145	1	Partial trisomy 13
GM3119	4	MC5A	20U 0172	28	F	WH	46,XX,t(9;17)(9pter>9p22::`7p11>17qter;17pter>17p11::9p22>9qter)	B	A	MOTHER	449	2	Fetus is GM3100 Amniotic
GM3196	3	HF12	20U 0318	31	F	WH	46,XX,t(17;22)(17qter>17p13::22q11>22qter;22pter>22q11::17p13>17pter),inv(5)(pter>p13::q13>p13::q13>qter)	B	A	PROBAND	518	1	See GM3197 Lymphoid; no hematological disorder; habitual aborter; normal phenotype
CHROMOSOME 18													
*GM0017	4	MC5A	20U 0012	30MO	M	BL	45,XY,-15,-18,+der(18),t(15;18)(18pter>18q23::15q1>15qter)	U	A	PROBAND			Multiple congenital anomalies & mental retardation
*GM0610	7	MC5A	20U 0075	11WK	F	WH	46,XX,t(6;18)(6pter>6q21::18p11>18pter;6qter>6q21::18p11>18qter)	B	A	PROBAND			
*GM0657	3	MC5A	20U 0107	4WK	M	WH	46,XY,t(7;18)(7pter>7q36::18q21>18qter;18pter>18q21::7q36>7qter)	B	A	PROBAND			
*GM1224	7	HF12	20U 0077	3MO	F	WH	46,XX,-13,+der(13),t(13;18)(13pter>13q34::18q11>18qter)pat	U	A	PROBAND			
GM1892	5	HF12	20U 0132	37	F	WH	46,XX,t(9;18)(9pter>9p24::18q12>18qter;18pter>18q12::9p24>9pter)	B	A	PROBAND			
GM2044	2	HF12	20U 0164	7	M		45,XY,-16,t(14;16)(16;18)(14qter>14p12::16p11>16pter;18qter>18p1::16q12>16qter)	U	A	PROBAND			
GM3089	6	MC5A	20U 0299	3WK	F	WH	46,XX,-18,+der(18),t(13;18)(13qter>13q31::18q23>18pter)pat	U	A	PROBAND	301	1	
CHROMOSOME 19													
*GM0089	5	MC5A	20U 0043		F	WH	46,X,t(X;19)(Xpter>Xq2::19q13 or 19p13>19qter or 19pter;19pter or 19qter>19p13 or 19q13::Xq2>Xqter)	B	A	PROBAND			
*GM0271	3	MC5A	20U 0058	28	F	WH	46,XX,t(17;19)(17pter>17q23::19p13>19pter;19qter>19p13::17q23>17qter)	B	A	PROBAND			Normal phenotype; 1 spontaneous abortion and 1 stillborn with anomalies
CHROMOSOME 20													
GM1225	10	HF12	20U 0077	3MO	F	IN	46,XX,t(2;20)(2qter>2p21::20p13>20pter;20qter>20p13::2p21>2pter)	B	A	PROBAND			Indian (India); de novo; has multiple congenital anomalies

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 20													
*GM0981	3	EEMEM	20U 0148	4	F	BL	46,XX,-14,+der(14),t(14;20)(14qter>14p11::20p11>20pter)mat,inv(9)(pter>p13::q13>p13::q13>qter)	U	A	PROBAND	146	1	
*GM0982	3	EEMEM	20U 0148		F	BL	46,XX,t(14;20)(14pter>14p11::20p11>20qter;20pter>20p11::14p11>14qter)	B	A	MOTHER	146	2	
CHROMOSOME 21													
*GM0098	5	MC5A	20U 0119	10MO	M		46,XY,-21,+der(21),t(4;21)(21qter>21p12::4p11>4pter)mat	U	A	PROBAND			
*GM0144	5	MC5A	20U 0012	34	F	WH	46,XX,-6,+der(6),t(6;21)(6qter>6p25::21q11>21qter)mat	U	A	PROBAND			13% of cells found to be balanced in passage 1 after recovery; these tend to increase with passage in culture
*GM0260	6	MC5A	20U 0095	11	M	WH	46,XY/47,XY,+21/47,XY,+t(21;21)(qter>cen>qter)	U	A	PROBAND	327	1	66%/24%/10% Mosaic; formerly GM0069
GM1564	9	EEMEM	20U 0151	28	F	WH	45,XX,t(1;21)(q42 or 43;q11)	U	B	PROBAND			Clinically normal
GM1665	3	HF12	20U 0140	28	F	WH	46,XX,t(12;21)(12qter>12p11::21p11>21pter;21qter>21p11::12p11>12pter)	B	A	PROBAND			
GM1700	3	HF12	20U 0095	6	M		46,XY,t(21;22)(21qter>21p1::22q11>22qter;22pter>22q11::21p1>21pter)	B	A	PROBAND			Mental retardation
GM1813	3	HF12	20U 0066	19	F	WH	46,XX,t(1;21)(q12;q22)pat/46,XX,t(1;21)(q12;q22)pat,t(X;15)(p11;q13)	U	A	PROBAND			Menstrual dysfunction; Adj-2 segregation; Mosaic 40%/60%
GM1881	7	MC5A	20U 0140	3	M	WH	46,XY,t(1;21)(1qter>1p31::21q22>21qter;21pter>21q22::1p31>1pter)	B	A	PROBAND			Sanfilippo A; deficient heparan N-sulfatase, normal alpha-N-acetylglucosaminidase activity
GM2058	5	HF12	20U 0165	11DA	F	WH	46,XX/46,XX,-21,+t(21;21)(21qter>cen>21qter)	U	A	PROBAND			87%/13% at passage 8
GM2524	7	MC5A	20U 0223	10WK	M	WH	47,XY,+21/46,XY,-21,+dic(21;21)(pter>q21::p12>cen>qter)	U	A	PROBAND			Mosaic; 82%/18%
GM3268	2	HF12	20U 0122	10MO	M	BL	46,XY,-21,+der(21),t(9;21)	U		PROBAND			Karyotype based upon lymphocytes
*GM0692	5	MC5A	20U 0001	8	M	WH	45,XY,-2,-21,+der(2),t(2;21)(2pter>2q37::21q2105>21qter)mat	U	A	PROBAND	133	1	Mentally retarded

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 21													
*GM0693	5	MC5A	20U 0001	27	F	WH	46,XX,t(2;21)(2pter>2q37::21q2105> 21qter;21pter>21q2105::2q37>2qter)	B	A	MOTHER	133	2	
*GM1399	5	HF12	20U 0132	20	M	BL	47,XY,+der(21),t(10;21)(21pter>21q2105:: 10q26>10qter)mat	U	A	MAT 1 COU	143	3	Mental retardation and ear anomalies; 1st cousin once removed
*GM1413	8	HF12	20U 0132	18MO	F	BL	46,XX,-10,+der(10),t(10;21)(10pter> 10q26::21q2105>21qter)mat	U	A	PROBAND	143	1	Down's syndrome
*GM1580	6	HF12	20U 0132	48	F	BL	46,XX,t(10;21)(10pter>10q26::21q21> 21qter)(21pter>21q21::10q26:10qter)	B	A	MAT GRMR	143	2	Normal phenotype
*GM0983	3	EEMEM	20U 0095	14MO	M	WH	46,XY,-21,+der(21),t(10;21)(10qter> 10q24::21q22>21pter)mat	U	A	PROBAND	144	1	
*GM0984	3	EEMEM	20U 0095	34	F	WH	46,XX,t(10;21)(10pter>10q24::21q22> 21qter;21pter>21q22::10q24>10qter)	B	A	MOTHER	144	2	
*GM1730	11	EEMEM	20U 0132	5	F	BL	46,X,-21,+der(21),t(X;21)(21qter>21p11:: Xq11>Xqter)mat	U	A	PROBAND	148	1	
CHROMOSOME 22													
*GM0005	5	MC5A	20U 0111	30	F	WH	45,XX,t(14;22)(14qter>cen>22qter)	U	A	PROBAND			
*GM0119	5	MC5A	20U 0087	31	F	WH	46,XX,t(17;22)(17qter>17p13::22q11> 22qter;22pter>22q11::17p13>17pter)	B	A	PROBAND			Clinically normal
*GM0392	9	MC5A	20U 0002	1	F	WH	45,XX,t(13;22)(13qter>cen>22qter)	U	A	PROBAND			
*GM0627	3	MC5A	20U 0058	30	F	BL	46,XX,t(13;22)(13pter>13q22::22q13> 22qter;22pter>22q13::13q22>13qter)	B	A	PROBAND			
*GM0980	2	MC5A	20U 0012	33	F	WH	45,XX,-11,-22,+der(11),t(11;22)(11pter> 11q25::22q11>22qter)	U	A	PROBAND			69% of cells also show t(1;4)(q25;p12)
GM1700	3	HF12	20U 0095	6	M		46,XY,t(21;22)(21qter>21p1::22q11> 22qter;22pter>22q11::21p1>21pter)	B	A	PROBAND			Mental retardation
GM2325	4	HF12	20U 0182	11DA	F	WH	47,XX,+der(22),t(16;22)(22pter>22q12:: 16p13>16pter)mat	U	A	PROBAND	179	1	Daughter of GM2324 Lymphoid
GM3196	3	HF12	20U 0318	31	F	WH	46,XX,t(17;22)(17qter>17p13::22q11> 22qter;22pter>22q11::17p13>17pter), inv(5)(pter>p13::q13>p13::q13>qter)	B	A	PROBAND	518	1	See GM3197 Lymphoid; no hematological disorder; habitual aborter; normal phenotype
GM3372	2	HF12	20U 0322	28	F	BL	46,XX,t(11;22)(11pter>11q25::22q13> 22qter;22pter>22q13::11q25>11qter)	B	A	MOTHER	613	2	Clinically normal

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 22													
GM3371	2	HF12	20U 0322	5WK	F	BL	47,XX,t(11;22)(q25;q13),+der(22),t(11;22)(22pter>22q13::11q25>11qter)mat,inv(9)(p13;q12)pat	U	A	PROBAND	613	1	Partial trisomy 22; father has same inv(9); developmental delay
							RECOMBINANT						
CHROMOSOME 01													
*GM2946	8	HF12	20U 0265	3MO	F	WH	46,XX,rec(1),dup q,inv ins(1)(pter>p22::q32>q31::p22>qter)	B	A	PROBAND			Partial trisomy 1
CHROMOSOME 03													
GM0366	3	MC5A	20U 0003	4	M	WH	46,XY,rec(3),dup q,inv(3)(qter>q21::p25>qter)	U	A	PROBAND	560	1	
GM3039	4	EEMEM	20U 0134	10MO	M	WH	46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)pat	U	A	PROBAND			Gross failure to thrive; multiple congenital anomalies
GM3085	9	EEMEM	20U 0224	17MO	M	WH	46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)pat	U	A	PROBAND			Multiple congenital anomalies
*GM1253	3	EEMEM	20U 0106	4MO	M	BL	46,XY,rec(3),dup q,inv(3)(qter>p25::q25>qter)mat	U	A	PROBAND	149	1	Son of GM1250 Fibroblast (XYY)
CHROMOSOME 13													
*GM1570	2	MC5A	20U 0119	16FW	M		46,XY,rec(13)dup p,inv(13)(pter>q22::p11>pter)pat	U	A	PROBAND	481	1	Paternal karyotype is 46,XY,inv(13)(p12q32)
							RING CHROMOSOME						
CHROMOSOME 06													
GM0109	7	MC5A	20U 0134	2	F	WH	46,XX,r(6)	U	A	PROBAND			12% r(6); 5% 46,XX; and 83% polyploid
CHROMOSOME 09													
*GM0166	9	MC5A	20U 0107	6	M	WH	45,XY,-9/46,XY,r(9)	U	A	PROBAND			44% of cells have r(9); 46% are 45,XY,-9
CHROMOSOME 13													
GM0285	3	MC5A	20U 0107	4DA	M	BL	46,XY,r(13)	B	A	PROBAND			Multiple congenital anomalies
*GM0729	8	MC5A	20U 0142	14	M	WH	45,XY,-13/46,XY,r(13)	U	A	PROBAND			Mental retardation
GM3321	2	HF12	20U 0066	11DA	F	WH	46,XX,r(13)	U	A	PROBAND			Greek; bilateral cleft lip & palate; microcephaly, microophthalmos; hypertelorism; 16% of cells have a fragment and no r(13)

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							RING CHROMOSOME						
CHROMOSOME 14													
GM3003	6	EEMEM	20U 0134	6	M	WH	46,XY,r(14)	U	A	PROBAND			Growth retardation; hypotonia; seizures
GM3233	2	HF12	20U 0122	1WK	M	WH	46,XY,r(14)/45,XY,-14	U	A	PROBAND			Congenital malformations; 88%/12%
CHROMOSOME 18													
GM1118	8	EEMEM	20U 0150	30MO	F	WH	46,XY,r(18)/45,XX,-18	U	A	PROBAND			78% of cells have r(18)
							INVERSION						
CHROMOSOME 01													
*GM2946	8	HF12	20U 0265	3MO	F	WH	46,XX,rec(1),dup q,inv ins(1)(pter>p22::q32>q31::p22>qter)	B	A	PROBAND			Partial trisomy 1
GM3126	2	MC5A	20U 0148	4	F	BL	46,XX,del inv(1)(pter>p13::q21>p13::q21>q42::)	U	A	PROBAND			Multiple congenital abnormalities; Tetrology of Fallot
CHROMOSOME 02													
GM0735	17	MC5A	20U 0077	17	F	WH	46,Xdic,Xqi,inv(2)(p15q21)mat	U	B	PROBAND			Turner's syndrome
CHROMOSOME 03													
GM0366	3	MC5A	20U 0003	4	M	WH	46,XY,rec(3),dup q,inv(3)(qter>q21::p25>qter)	U	A	PROBAND	560	1	
GM3039	4	EEMEM	20U 0134	10MO	M	WH	46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)pat	U	A	PROBAND			Gross failure to thrive; multiple congenital anomalies
GM3085	9	EEMEM	20U 0224	17MO	M	WH	46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)pat	U	A	PROBAND			Multiple congenital anomalies
*GM1252	3	EEMEM	20U 0106	22	F	BL	46,XX,inv(3)(pter>p25::q25>p25::q25>qter),inv(9)(pter>p11::q1>p11::q1>qter)	B	A	MOTHER	149	2	
*GM1253	3	EEMEM	20U 0106	4MO	M	BL	46,XY,rec(3),dup q,inv(3)(qter>p25::q25>qter)mat	U	A	PROBAND	149	1	Son of GM1250 Fibroblast (XYY)
CHROMOSOME 04													
GM0413	4	HF12	20U 0056	1	M		46,XY,inv(4)(pter>p14::q2>p14::q2>qter)mat	B	A	PROBAND	150	1	Minor malformations
GM0414	3	HF12	20U 0056	24	F		46,XX,inv(4)(pter>p14::q2>p14::q2>qter)	B	A	MOTHER	150	2	Clinically normal

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							INVERSION						
CHROMOSOME 05													
*GM2137	2	HF12	20U 0012	2	F	WH	47,XX,inv(5)(pter>p13::q13>p13::q13>qter)mat,+mar	U	A	PROBAND	453	1	Marker may be rec(5)(p1q1)mat
GM2335	3	HF12	20U 0125	21	M	WH	46,XY,inv(5)(pter>p1505::q12>p1505::q12>qter)	B	A	PROBAND	217	1	G6PD(B)+ Xg(a) antigen absent; parents are GM2334 and GM2336 Fibroblast
GM3196	3	HF12	20U 0318	31	F	WH	46,XX,inv(5)(pter>p13::q13>p13::q13>qter),t(17;22)(17qter>17p13::22q11>22qter;22pter>22q11::17p13>17pter)	B	A	PROBAND	518	1	See GM3197 Lymphoid; no hematological disorder; habitual aborter; normal phenotype
CHROMOSOME 06													
GM2957	4	HF12	20U 0315	23	F	WH	46,XX,inv(6)(pter>p23::q21>p23::q21>qter)	B	A	PROBAND			
CHROMOSOME 09													
GM0425	12	R1640	20U 0013	10FW	M		47,XY,+8,inv(9)(pter>p11::q21>p11::q21>qter)	U	A	PROBAND			
*GM0981	3	EEMEM	20U 0148	4	F	BL	46,XX,-14,+der(14),t(14;20)(14qter>14p11::20p11>20pter)mat,inv(9)(pter>p13::q13>p13::q13>qter)	U	A	PROBAND	146	1	
*GM1251	2	EEMEM	20U 0106	4	M	BL	46,XY,inv(9)(pter>p11::q1>p11::q1>qter)mat	B	A	BRO	149	4	Clinically normal; son of GM1250 Fibroblast; brother of GM1253 Fibroblast
*GM1252	3	EEMEM	20U 0106	22	F	BL	46,XX,inv(3)(pter>p25::q25>p25::q25>qter),inv(9)(pter>p11::q1>p11::q1>qter)	B	A	MOTHER	149	2	
GM0445	7	MC5A	20U 0030	26	M	WH	46,XY,inv(9)(p13q13)	B	B	FATHER	151	2	Clinically normal
GM0447	7	MC5A	20U 0030	1	M	WH	46,XY,inv(9)(p13q13)	B	B	PROBAND	151	1	Congenital malformations
GM0450	7	MC5A	20U 0030	33	M	WH	46,XY,inv(9)(p13q13)	B	B	PROBAND	152	1	Clinically normal; skin biopsy
GM0451	7	MC5A	20U 0030	33	M	WH	46,XY,inv(9)(p13q13)	B	B	PROBAND	152	1	Same patient as GM0450; testicular tissue fibroblast culture
*GM1920	2	HF12	20U 0061	23	M	WH	47,XY,+21,inv(9)(pter>p13::q13>p13::q13>qter)mat	U	A	PROBAND	418	1	See GM1921 Lymphoid
GM3120	5	MC5A	20U 0172	5	F		45,XO,inv(9)(p11q13)/46,XY,inv(9)(p11q13)	U	B	PROBAND	450	1	Cultures from left gonad (GM3121) and right gonad (GM3122) also available; 90%/10%

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							INVERSION						
CHROMOSOME 09													
GM3371	2	HF12	20U 0322	5WK	F	BL	47,XX,t(11;22)(q25;q13),+der(22),t(11;22)(22pter>22q13::11q25>11qter)mat,inv(9)(p13;q12)pat	U	A	PROBAND	613	1	Partial trisomy 22; father has same inv(9); developmental delay
CHROMOSOME 10													
GM0448	7	MC5A	20U 0030	1	M	WH	46,XY,inv(10)(pter>p11::q21>p11::q21>qter)pat	B	A	PROBAND	153	1	Congenital malformations
GM0452	7	MC5A	20U 0030	22	M	WH	46,XY,inv(10)(pter>p11::q21>p11::q21>qter)	B	A	FATHER	153	2	Clinically normal
CHROMOSOME 13													
GM0437	3	MC5A	20U 0066	32	F	WH	46,XX,inv(13)(qter>q21::p1>q21)	B	A	PROBAND			
*GM1570	2	MC5A	20U 0119	16FW	M		46,XY,rec(13)dup p,inv(13)(pter>q22::p11>pter)pat	U	A	PROBAND	481	1	Paternal karyotype is 46,XY,inv(13)(p12q32)
CHROMOSOME 15													
GM3666	4	HF12	20U 0326	34	M		46,XY,+inv dup(15)	U	B	PROBAND			Clinically normal
							INSERTION						
CHROMOSOME 01													
GM2836	4	HF12	20U 0263	43	M	WH	46,XY,t(9;17)(q12;p11)/46,XY,t(9;17)(q12;p11),ins(3;1)(q21;q25q44)	B	A	PROBAND			61%/29%
*GM2946	8	HF12	20U 0265	3MO	F	WH	46,XX,rec(1),dup q,inv ins(1)(pter>p22::q32>q31::p22>qter)	B	A	PROBAND			Partial trisomy 1
GM0860	11	HF12	20U 0036	13	F	WH	46,XX,-5,+der(5),ins(5;1)(5pter>5q15::1q25>1q32::5q15>5qter)pat	U	A	PROBAND	128	1	Culture grows slowly
GM0861	7	HF12	20U 0036	40	M	WH	46,XY,ins(5;1)(5pter>5q15::1q25>1q32::5q15>5qter;1pter>1q25::1q32>1qter)	B	A	FATHER	128	2	
CHROMOSOME 03													
GM2486	3	MC5A	20U 0140	5MO	F		46,XX,ins(3)(pter>q21::q21>q27::q21>qter)	U	A	PROBAND			
GM2836	4	HF12	20U 0263	43	M	WH	46,XY,t(9;17)(q12;p11)/46,XY,t(9;17)(q12;p11),ins(3;1)(q21;q25q44)	B	A	PROBAND			61%/29%
CHROMOSOME 05													
GM0860	11	HF12	20U 0036	13	F	WH	46,XX,-5,+der(5),ins(5;1)(5pter>5q15::1q25::1q32>::5q15>5qter)pat	U	A	PROBAND	128	1	Culture grows slowly

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							INSERTION						
CHROMOSOME 05													
GM0861	7	HF12	20U	0036	40	M	WH 46,XY,ins(5;1)(5pter>5q15::1q25>1q32::5q15>5qter;1pter>1q25::1q32>1qter)	B	A	FATHER	128	2	
*GM1221	6	EEMEM	20U	0021	18MO	M	BL 46,XY,-5,+der(5),ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter)mat	U	A	PROBAND	138	1	Partial trisomy of 6q15q27
*GM1222	5	EEMEM	20U	0021	23	F	BL 46,XX,ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter;6pter>6q15::6q27>6qter)	B	A	MOTHER	138	2	
*GM1524	3	EEMEM	20U	0021	19	M	BL 46,XY,-5,+der(5),ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter)	U	A	1ST COU	138	3	
CHROMOSOME 06													
*GM1221	6	EEMEM	20U	0021	18MO	M	BL 46,XY,-5,+der(5),ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter)mat	U	A	PROBAND	138	1	Partial trisomy of 6q15q27
*GM1222	5	EEMEM	20U	0021	23	F	BL 46,XX,ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter;6pter>6q15::6q27>6qter)	B	A	MOTHER	138	2	
*GM1524	3	EEMEM	20U	0021	19	M	BL 46,XY,-5,+der(5),ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter)	U	A	1ST COU	138	3	
							ISOCHROMOSOME						
CHROMOSOME X													
*GM0088	5	MC5A	20U	0012	19	F	WH 46,X,i(X)(qter>cen>qter)	U	A	PROBAND			Buccal smear=18% sex chromatin +; Turner's syndrome stigmata
GM0339	3	MC5A	20U	0030	25	F	WH 45,X/46,X,dic,i(X)(qter>p11::p11>qter)	U	A	PROBAND			60%/40% Mosaic
GM0735	17	MC5A	20U	0077	17	F	WH 46,Xqi,Xdic,inv(2)(p15q21)mat	U	B	PROBAND			Turner's syndrome
GM2595	6	HF12	20U	0073	33	F	WH 46,X,i(X)(qter>cen>qter)	U	A	PROBAND			Turner's syndrome
CHROMOSOME Y													
GM1709	4	EEMEM	20U	0109	11	F	WH 46,X,i(Yq)	U	B	PROBAND			Gonadal dysgenesis
GM3774	2	HF12	20U	0315	16	F	45,XO/46,X isoY(q)	U	B	PROBAND			Turner mosaic; 65%/35%
CHROMOSOME 18													
GM3767	2	HF12	20U	0204	3	M	WH 46,XY,del(18)(p11)/46,XY,i(18q)	U	B	PROBAND			Clinical features of 18p-; lymphocytes show 70% 18p- and 30% i(18q)

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DUPLICATION						
CHROMOSOME 01													
*GM2946	8	HF12	20U	0265	3MO	F	WH 46,XX,rec(1),dup q,inv ins(1)(pter>p22::q32>q31::p22>qter)	B	A	PROBAND			Partial trisomy 1
CHROMOSOME 02													
GM2848	3	MC5A	20U	0070	4	M	WH 46,XY,-2,+dup(2)(2qter>2q33::2p25>2qter)	U	A	PROBAND			In lymphocytes, 28% of the cells have r(2)(2p25>2q33) instead of the duplication
CHROMOSOME 03													
GM0366	3	MC5A	20U	0003	4	M	WH 46,XY,rec(3),dup q,inv(3)(qter>q21::p25>qter)	U	A	PROBAND	560	1	
GM3039	4	EEMEM	20U	0134	10MO	M	WH 46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)pat	U	A	PROBAND			Gross failure to thrive; multiple congenital anomalies
GM3085	9	EEMEM	20U	0224	17MO	M	WH 46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)pat	U	A	PROBAND			Multiple congenital anomalies
*GM1253	3	EEMEM	20U	0106	4MO	M	BL 46,XY,rec(3),dup q,inv(3)(qter>p25::q25>qter)mat	U	A	PROBAND	149	1	Son of GM1250 Fibroblast (XYY)
CHROMOSOME 13													
*GM1570	2	MC5A	20U	0119	16FW	M	46,XY,rec(13)dup p,inv(13)(pter>q22::p11>pter)pat	U	A	PROBAND	481	1	Paternal karyotype is 46,XY,inv(13)(p12q32)
CHROMOSOME 15													
GM3666	4	HF12	20U	0326	34	M	46,XY,+inv dup(15)	U	B	PROBAND			Clinically normal
							DICENTRIC CHROMOSOME						
CHROMOSOME X													
GM0314	16	MC5A	20U	0030	60	F	WH 45,X/46,X,dic(X)(qter>p22::p22>qter)	U	A	PROBAND			Ovarian dysgenesis without Turner stigmata; 70% of cells have the dicentric chromosome
GM0339	3	MC5A	20U	0030	25	F	WH 45,X/46,X,dic,i(X)(qter>p11::p11>qter)	U	A	PROBAND			60%/40% Mosaic
GM0735	17	MC5A	20U	0077	17	F	WH 46,Xqi,Xdic,inv(2)(p15q21)mat	U	B	PROBAND			Turner's syndrome
CHROMOSOME 13													
GM3518		HF12	20U	0265	4MO	F	BL 46,XX,dic(13pter>13q34::13q34>13pter)	U	B	PROBAND			Severe microcephaly, oligodactyly, coloboma, midline scalp defect, hypoplastic nose

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DICENTRIC CHROMOSOME						
CHROMOSOME 21													
GM2524	7	MC5A	20U 0223	10WK	M	WH	47,XY,+21/46,XY,-21,+dic(21;21)(pter>q21::p12>cen>qter)	U	A	PROBAND			Mosaic; 82%/18%
							DERIVATIVE CHROMOSOME						
CHROMOSOME 01													
GM1229	3	HF12	20U 0007	3	F	BL	46,XX,-1,+der(1),t(1;2)(1qter>1p36::2q31>2qter)pat	U	A	PROBAND	129	1	Multiple congenital anomalies & mental retardation
GM3369	3	HF12	20U 0322	23	F	WH	46,XX,-1,+der(1),t(1;10)(1pter>1q44::10p11>10pter)mat	U	A	PROBAND	612	1	Developmental delay
CHROMOSOME 02													
*GM0501	3	MC5A	20U 0014	5	M	WH	46,XY,-2,+der(2),t(2;4)(2qter>2p25::4q21>4qter)pat	U	A	PROBAND	131	1	
CHROMOSOME 04													
GM1183	4	HF12	20U 0007	NB	M	WH	46,XY,-4,+der(4),t(4;12)(4qter>4p1608::12p11>12pter)pat	U	A	PROBAND			
*GM1220	3	MC5A	20U 0066	2	F		46,XX,-4,+der(4),t(4;7)(4qter>4p16::7q34>7qter)mat	U	A	PROBAND	136	1	
CHROMOSOME 05													
*GM0071	15	MC5A	20U 0015	19	F	WH	46,XX,-5,der(5),t(5;?)(p1;?)mat	U	A	PROBAND			Cri du Chat; deletion assumed to be derived rather than a simple deletion; ATCC CCL 90
*GM0344	4	MC5A	20U 0066	7	M	WH	45,XY,-5,-15,+der(5),t(5;15)(5qter>5p14::15q11>15qter),t(9;11)(9qter>9p24::11q23>11qter;11pter>11q23::9p24>9pter)	U	A	PROBAND			Cri du Chat
GM0860	11	HF12	20U 0036	13	F	WH	46,XX,-5,+der(5),ins(5;1)(5pter>5q15::1q25>1q32::5q15>5qter)pat	U	A	PROBAND	128	1	Culture grows slowly
GM3493	2	HF12	20U 0204	5MO	F	WH	46,XX,-5,+der(5),t(5;6)(5pter>5q35::6p22>6pter)pat	U	A	PROBAND	135	1	Partial trisomy 6p; psychomotor retardation; multiple congenital anomalies
*GM1524	3	EEMEM	20U 0021	19	M	BL	46,XY,-5,+der(5),ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter)	U	A	1ST COU	138	3	
CHROMOSOME 06													
*GM0144	5	MC5A	20U 0012	34	F	WH	46,XX,-6,+der(6),t(6;21)(6qter>6p25::21q11>21qter)mat	U	A	PROBAND			13% of cells found to be balanced in passage 1 after recovery; these tend to increase with passage in culture

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DERIVATIVE CHROMOSOME						
CHROMOSOME 06													
*GM3647	2	HF12	20U 0204	41	M	WH	46,XY,-6,+der(6),t(1;6)(6qter>6p25::1q32>1qter)mat	U	A	PROBAND	297	1	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1
*GM3648	2	HF12	20U 0204	32	M	WH	46,XY,-6,+der(6),t(1;6)(6qter>6p25::1q32>1qter)mat	U	A	HALF BRO	297	2	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1
CHROMOSOME 09													
*GM1387	4	MC5A	20U 0061	14	F	WH	46,XX,-13,+der(9),t(9;13)(q22;q12)mat	U	B	PROBAND	369	1	Result of Adj-2 segregation; see also GM1388 Lymphoid
GM3563	2	MC5A	20U 0148	1MO	M	WH	46,XY,der(9),t(3;9)(q12;p24)mat	U	B	PROBAND			Multiple congenital anomalies
*GM1414	5	EEMEM	20U 0068	28	F	BL	46,X,+der(9),t(X;9)(9pter>9q34::Xq13>Xqter)mat	U	A	PROBAND	140	1	Turner's syndrome
CHROMOSOME 10													
*GM1396	3	MC5A	20U 0095	3	M	WH	46,XY,-10,+der(10),t(10;16)(10pter>10q26::16q22>16qter)pat	U	A	PROBAND			
*GM3689	6	EEMEM	20U 0247	24	F	WH	46,XX,-10,+der(10),t(8;10)(8pter>8p21.1::10p15>10qter)mat	U	A	PROBAND			Mental & growth retardation; delayed sexual maturation; multiple congenital anomalies
GM1848	3	HF12	20U 0179	35	M	WH	46,XY,-10,+der(10),t(2;10)(10pter>10q26::2p24>2pter)mat	U	A	MAT 1 COU	134	2	Possible red cell acid phosphatase variant
GM1849	4	HF12	20U 0179	26	M	WH	46,XY,der(10),t(2;10)(p25;q26)mat	U	B	MAT 1 COU	134	3	Possible red cell acid phosphatase variant
*GM1413	8	HF12	20U 0132	18MO	F	BL	46,XX,-10,+der(10),t(10;21)(10pter>10q26::21q2105>21qter)mat	U	A	PROBAND	143	1	Down's syndrome
CHROMOSOME 11													
*GM0980	2	MC5A	20U 0012	33	F	WH	45,XX,-11,-22,+der(11),t(11;22)(11pter>11q25::22q11>22qter)	U	A	PROBAND			69% of cells also show t(1;4)(q25;p12)

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DERIVATIVE CHROMOSOME						
CHROMOSOME 12													
GM1535	6	MC5A	20U 0002	16	F	BL	46,XX,-12,+der(12),t(5;12)(12pter> 12q24::5q33>5qter)mat	U	A	PROBAND	139	1	
GM2819	3	HF12	20U 0107	7MO	F	BL	46,XX,-12,+der(12),t(9;12)(12pter> 12q24::9p13>9pter)mat	U	A	PROBAND	324	1	See GM2820 Lymphoid
CHROMOSOME 13													
*GM1224	7	HF12	20U 0077	3MO	F	WH	46,XX,-13,+der(13),t(13;18)(13pter> 13q34::18q11>18qter)pat	U	A	PROBAND			
GM2813	7	R1640	20U 0148	1WK	F	WH	46,XX,-15,+der(13q15q),t(13;15)(13qter> cen>15qter)mat	U	A	PROBAND			
*GM1555	5	EEMEM	20U 0113	13	M	WH	47,XY,+der(13),t(13;17)(13pter>13q14:: 17p13>17pter)mat	U	A	BRO	145	2	
CHROMOSOME 14													
*GM0981	3	EEMEM	20U 0148	4	F	BL	46,XX,-14,+der(14),t(14;20)(14qter> 14p11::20p11>20pter)mat,inv(9)(pter> p13::q13>p13::q13>qter)	U	A	PROBAND	146	1	
*GM0074	10	HEMEM	10U 0111	23	M	WH	47,XY,t(X;14)(Xpter>Xq13::14q32>14qter; 14pter>14q32::Xq13>Xqter)+der(14), t(X;14)(14pter>14q32::Xq13>Xqter)mat	U	A	PROBAND	147	1	Klinefelter's syndrome
GM3462	2	HF12	20U 0204	3MO	F	WH	47,XX,+der(14),t(9;14)(14pter>14q13:: 9p22>9pter)mat	U	A	PROBAND	227	1	Growth retardation and congenital malformations
GM1750	2	HF12	20U 0070	30MO	M	WH	47,XY,+der(14),t(9;14)(p24;q22)mat	U	B	PROBAND	141	1	
CHROMOSOME 15													
*GM0118	5	MC5A	20U 0052	29	F	WH	46,XX,-15,+der(15),t(Y;15)(Yqter>Yq11:: 15p1>15qter)pat	U	A	PROBAND			Uterine tissue fibroblast culture
GM1734	3	HF12	20U 0140	40	M		46,XY,der(15),t(9;15)(p11;q11)mat	U	B	PROBAND	257	1	
GM2813	7	R1640	20U 0148	1WK	F	WH	46,XX,-15,+der(13q15q),t(13;15)(13qter> cen>15qter)mat	U	A	PROBAND			
GM2488		MC5A	20U 0140	23	M	WH	46,XY,-15,+der(15),t(5;15)(5pter>5p13:: 15p11>15qter)mat	U	A	PROBAND	225	1	Passage 6 at IMR
GM3255	3	HF12	20U 0204	5	M	WH	46,XY,-15,+der(15),t(8;15)(15pter> 15q26::8p21>8pter)mat	U	A	PROBAND	609	1	Partial trisomy 8(p);
CHROMOSOME 17													
*GM0217	6	MC5A	20U 0143	9DA	M	WH	46,XY,-17,+der(17),t(10;17)(17qter> 17p13::10q24>10qter)pat	U	A	PROBAND	142	1	Culture grows slowly

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DERIVATIVE CHROMOSOME						
CHROMOSOME 17													
*GM0959	6	EEMEM	20U 0143	11	M	WH	46,XY,-17,+der(17),t(10;17)(17qter>17p13::10q24>10qter)pat	U	A	MAT 3RD COU	142	3	
*GM1663	13	MC5A	20U 0113	14	M	WH	46,XY,-17,+der(17),t(13;17)(17qter>17p13::13q14>13qter)mat	U	A	PROBAND	145	1	Partial trisomy 13
CHROMOSOME 21													
*GM0098	5	MC5A	20U 0119	10MO	M		46,XY,-21,+der(21),t(4;21)(21qter>21p12::4p11>4pter)mat	U	A	PROBAND			
GM3268	2	HF12	20U 0122	10MO	M	BL	46,XY,-21,+der(21),t(9;21)	U		PROBAND			Karyotype based upon lymphocytes
*GM1399	5	HF12	20U 0132	20	M	BL	47,XY,+der(21),t(10;21)(21pter>21q2105::10q26>10qter)mat	U	A	MAT 1 COU	143	3	Mental retardation and ear anomalies; 1st cousin once removed
*GM0983	3	EEMEM	20U 0095	14MO	M	WH	46,XY,-21,+der(21),t(10;21)(10qter>10q24::21q22>21pter)mat	U	A	PROBAND	144	1	
*GM1730	11	EEMEM	20U 0132	5	F	BL	46,X,-21,+der(21),t(X;21)(21qter>21p11::Xq11>Xqter)mat	U	A	PROBAND	148	1	
CHROMOSOME 22													
GM3298	3	HF12	20U 0016	8DA	M	WH	47,XY,+der(22)mat	U		PROBAND			Karyotype based upon lymphocytes
GM3371	2	HF12	20U 0322	5WK	F	BL	47,XX,t(11;22)(q25;q13),+der(22),t(11;22)(22pter>22q13::11q25>11qter)mat,inv(9)(p13;q12)pat	U	A	PROBAND	613	1	Partial trisomy 22; father has same inv(9); developmental delay
							DELETION						
CHROMOSOME X													
GM1941	3	HF12	20U 0012	13	F	WH	45,X/46,X,del(X)(q11)	U		PROBAND			Karyotype based upon peripheral blood leukocytes
*GM2470	9	HF12	20U 0030	55	F	WH	46,X,del(X)(pter>p22::p11>qter)	U	A	PROBAND			
GM3624	6	EEMEM	20U 0204	20	F	WH	46,X,del(X)(q24)	U	B	PROBAND			Phenotypically normal; normal fertility
CHROMOSOME Y													
GM2668	4	HF12	20U 0229	41	A	WH	45,X/46,X,del(Y)(pter>q12:)	U	A	PROBAND			Multiple congenital anomalies; 33%/66%
GM2730	10	EEMEM	20U 0109	4DA	A	WH	45,X/46,X,del(Y)(pter>q11:)	U	A	PROBAND			Breakpoint is above heterochromatic region; 33%/66%

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
DELETION													
CHROMOSOME 01													
*GM0214	8	MC5A	20U 0030	2	M	WH	46,XY,del(1)(pter>q25::q32>qter)	U	A	PROBAND			Growth hormone deficiency and hypothyroidism
GM0803	3	EEMEM	20U 0002	2	M	WH	46,XY,del(1)(q24>q25)	U		PROBAND			Karyotype based on lymphocytes
*GM2025	4	HF12	20U 0126	30MO	M	WH	46,XY,del(1)(pter>q21 or q22::q25>qter)	U	A	PROBAND			Multiple congenital anomalies
GM2906A	8	HF12	20U 0014	3WK	F	WH	46,XX,del(1)(pter>q23::q32>qter)	U	A	PROBAND			Bilateral cleft lip & palate; brachydactyly and clinodactyly
GM3126	2	MC5A	20U 0148	4	F	BL	46,XX,del inv(1)(pter>p13::q21>p13::q21>q42:)	U	A	PROBAND			Multiple congenital abnormalities; Tetrology of Fallot
CHROMOSOME 02													
*GM0945	2	EEMEM	20U 0077	19MO	F	OR	46,XX,del(2)(qter>p24:)	U	A	PROBAND			Failure to thrive; severe retardation
*GM1138	3	MC5A	20U 0095	1	M	BL	46,XY,del(2)(qter>p23:)	U	A	PROBAND			
CHROMOSOME 04													
*GM0072	15	MC5A	20U 0015	11	M	WH	46,XY,del(4)(qter>p14:)	U	A	PROBAND			Wolf's syndrome
GM2003	4	HF12	20U 0095	11	F	WH	46,XX,del(4)(pter>q33:)	U	A	PROBAND			
GM3013	3	MC5A	20U 0148	1MO	F	WH	46,XX,del(4)(pter>q313:)	U	A	PROBAND			Multiple congenital anomalies
GM0343	3	MC5A	20U 0066	3	M		46,XY,del(4)(qter>p14:)	U	A	PROBAND	347	1	Wolf-Hirschhorn syndrome
CHROMOSOME 05													
*GM0071	15	MC5A	20U 0015	19	F	WH	46,XX,-5,der(5),t(5;?)(p1;?)mat	U	A	PROBAND			Cri du Chat; deletion assumed to be derived rather than a simple deletion; ATCC CCL 90
GM2337	3	HF12	20U 0066	35	F	WH	46,XX,del(5)(qter>p1403:)	U	A	PROBAND			Cri du Chat
GM2800	4	MC5A	20U 0148	3WK	F	BL	46,XX,del(5)(qter>p1405:)	U	A	PROBAND			Not Cri du Chat syndrome

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DELETION						
CHROMOSOME 07													
*GM1059	5	HF12	10U 0149	50DA	F	WH	46,XX,del(7)(pter>q22::q32>qter)	U	A	PROBAND			Hypertonia, seizures, feeding difficulty, & multiple congenital anomalies
GM3240	8	HF12	20U 0016	7DA	M	WH	46,XY,del(7)(7pter>7q34:)	U	A	PROBAND			Microcephaly & other congenital anomalies
GM3733	2	HF12	20U 0204	7	M	WH	46,XY,del(7)(q32)	U		PROBAND			Motor retardation; mild dysmorphic features; karyotype based upon lymphocytes
CHROMOSOME 08													
GM1549	8	MC5A	20U 0140	7	M		46,XY,del(8)(qter>p21:)	U	A	PROBAND			Multiple congenital anomalies & mental retardation
*GM3134	2	HF12	20U 0204	3MO	M	WH	46,XY,del(8)(pter>q13::q22>qter)	U	A	PROBAND			Multiple congenital anomalies
CHROMOSOME 09													
*GM0870	2	EEMEM	20U 0015	22	F	WH	46,XX,del(9)(qter>p13:)	U	A	PROBAND			Severe mental retardation & congenital anomalies
GM1667	7	EEMEM	20U 0002	13	F	WH	46,XX,del(9)(p22)	U	B	PROBAND			
*GM1893	4	HF12	20U 0140	3	M		46,XY,del(9)(pter>q11::q2200>qter)	U	A	PROBAND			Mental retardation, multiple anomalies, seizures
GM2356	2	HF12	20U 0140	3	M		46,XY,del(9)(9qter>9p22:)	U	A	PROBAND			Dysplasia
CHROMOSOME 10													
GM3047	7	HF12	20U 0016	5	M	WH	46,XY,del(10)(qter>p11:)	U	A	PROBAND			Psychomotor retardation, trigonencephaly, and multiple congenital anomalies
CHROMOSOME 11													
GM2008	6	HF12	20U 0069	2WK	F	BL	46,XX,del(11)(pter>q24:)	U	A	PROBAND			Multiple congenital anomalies & bilateral colobomata
CHROMOSOME 13													
*GM0509	4	MC5A	20U 0143	16MO	F	BL	46,XX,del(13)(pter>q14:)	U	A	PROBAND	345	1	Short term lymphocytes show r(13); formerly GM0250

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DELETION						
CHROMOSOME 16													
*GM2346	2	HF12	20U	0204	3MO	M	WH 46,XY,del(16)(pter>q22:)	U	A	PROBAND			Hydrocephalus, hypospadius, & minor dysmorphic features
CHROMOSOME 18													
GM1727	13	MC5A	20U	0142	45	M	WH 46,XY,del(18)(qter>p11:)	U	A	PROBAND			Mentally retarded; aberrations involving chromosomes 1, 10, 15, & 17 were also found in all cells examined
GM2980	2	HF12	20U	0107	12	M	WH 46,XY,del(18)(pter>q2108:)	U	A	PROBAND			
GM3767	2	HF12	20U	0204	3	M	WH 46,XY,del(18)(p11)/46,XY,i(18q)	U	B	PROBAND			Clinical features of 18p-; lymphocytes show 70% 18p- and 30% i(18q)
							TRIPLOID						
CHROMOSOME 3N													
*GM1322	3	MC5A	20U	0095	2DA	F	BL 69,XXY	U	A	PROBAND			Meningomyelocele
GM1672	8	EEMEM	10U	0088	1DA	M	WH 69,XXY	U	A	PROBAND			
GM3591	6	HF12	20U	0107	26FW	M	WH 69,XXY	U	B	PROBAND			Multiple anomalies
							ANEUPLOID						
CHROMOSOME X													
*GM0157	12	MC5A	20U	0030	28	M	WH 49,XXXXY,t(4;11)(4pter>4q3::11q23> 11qter;11pter>11q23::4q3>4qter)	U	A	PROBAND			Severe mental retardation; sexual infantilism
GM0254	8	MC5A	20U	0075	10FW	F	BL 47,XXX	U	A	PROBAND			76% interphase nuclei have 2 sex chromosome bodies; 15% have one; no evidence for mosaicism
GM0314	16	MC5A	20U	0030	60	F	WH 45,X/46,X,dic(X)(qter>p22::p22>qter)	U	A	PROBAND			Ovarian dysgenesis without Turner stigmata; 70% of cells have the dicentric chromosome
GM0324	5	MC5A	20U	0125	22	M	WH 47,XXY	U		PROBAND			Sardinian; homozygous for G6PD (med. type), Xg(a) antigen present

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							ANEUPLOID						
CHROMOSOME X													
GM0325	4	MC5A	20U 0125	30	M	WH	47,XXY	U		PROBAND			Sardinian; heterozygous for G6PD deficiency (med. type), Xg(a) antigen present
GM0326	3	MC5A	20U 0125	6	M	WH	49,XXXXY	U	A	PROBAND	164	4	Normal RBC G6PD level; Xg(a) antigen positive; severe retardation; hypogonadism; parents are GM0321 & GM0323 Fibroblast
GM0775	4	EEMEM	20U 0107	13	F		45,X	U	A	PROBAND			Turner's syndrome
GM0857	3	MC5A	20U 0095	1DA	F	WH	45,X	U	A	PROBAND			Turner's syndrome
GM0993	2	MC5A	20U 0107	9	F	BL	45,X	U	A	PROBAND			Turner's syndrome
*GM1137	6	R1640	10U 0017	1MO	M	BL	48,XXY,+21,t(6;10)(6qter>6p22 or 24::10p12>10pter;10qter>10p12::6p22 or 24>6pter)	U	B	PROBAND			Down's syndrome features
GM1176	2	MC5A	20U 0107	8	F	WH	45,X	U	A	PROBAND			Turner's syndrome
*GM1415	3	MC5A	20U 0061	27	F	WH	48,XXXX	U	A	PROBAND	370	1	4% polyploidy; see GM1416 Lymphoid
GM1441	2	MC5A	20U 0074	3MO	M	WH	45,X	U	A	PROBAND			Tissue from right testes; lymphocytes showed 45,X/46,X,iso(Y); 80%/20%
GM1723	2	HF12	20U 0203	23	F	WH	45,X	U	A	PROBAND	394	1	Skin biopsy; fibroblast culture from uterine tissue also available (GM1724)
*GM2521	9	EEMEM	15U 0222	24	M	WH	48,XXYY	U	A	PROBAND			High levels of gonadotrophins
GM3102	5	MC5A	20U 0172	18FW	M	WH	47,XXY	U	A	PROBAND	448	1	See GM3091 Amniotic; Klinefelter's
GM3133	6	HF12	20U 0273	FW	F		46,X,+15	U	A	PROBAND			
GM3623	6	MC5A	20U 0340	NB	F	BL	48,XXX,+18	U	A	PROBAND			

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							ANEUPLOID						
CHROMOSOME X													
GM0562	4	MC5A	20U 0095	14	F	BL	45,X/46,XX	U	A	PROBAND	598	1	4 cells 46,XX, 46 cells 45,XO; tissue from left gonad; gonad revealed testicular rather than ovarian tissue
GM0563	4	MC5A	20U 0095	14	F	BL	45,X	U	A	SAME	598	1	Only XO cells found; tissue from right gonad; gonad revealed testicular rather than ovarian tissue
CHROMOSOME Y													
GM0978	7	EEMEM	20U 0143	15	F	WH	45,X/46,XY	U	A	PROBAND	334	1	Formerly GM0188; 97%/3%
*GM2521	9	EEMEM	15U 0222	24	M	WH	48,XXYY	U	A	PROBAND			High levels of gonadotrophins
GM3495	6	EEMEM	20U 0332	15MO	M	WH	49,XYYYY	U	B	PROBAND			Hypertelorism; low set ears; abnormal dermatoglyphics; clinodactyly
GM3774	2	HF12	20U 0315	16	F		45,XO/46,X isoY(q)	U	B	PROBAND			Turner mosaic; 65%/35%
GM1250	3	EEMEM	20U 0106	23	M	BL	47,XYY	U	A	FATHER	149	3	See GM1253 Fibroblast (son)
CHROMOSOME 02													
GM3576	10	MC5A	20U 0326	13FW	M	WH	48,XY,+2,+21	U	A	PROBAND			
CHROMOSOME 08													
GM0425	12	R1640	20U 0013	10FW	M		47,XY,+8,inv(9)(pter>p11::q21>p11::q21>qter)	U	A	PROBAND			
*GM0496	3	MC5A	20U 0056	2	M		46,XY,/47,XY,+8	U	A	PROBAND			2 cell lines in lymphocytes, 83% have +8; in skin fibroblasts 81% have +8
*GM2030	7	MC5A	20U 0030	23	M	WH	46,XY/47,XY,+8	U	A	PROBAND			In fibroblasts 58% have +8; mental retardation
GM2596	6	EEMEM	20U 0073	10MO	F	BL	46,XX/47,XX,+8	U	A	PROBAND			58% trisomy 8; mental retardation & failure to thrive
CHROMOSOME 09													
*GM2329	7	EEMEM	10U 0039	18DA	F	WH	46,XX/47,XX,+9	U	A	PROBAND			10% of fibroblasts show the +9

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							ANEUPLOID						
CHROMOSOME 09													
GM3225	4	HF12	20U 0081	31	F	BL	46,XX/47,XX,+9p	U	A	MOTHER	602	2	36%/64% in lymphocytes, 98%/2% in fibroblasts
GM3226	2	HF12	20U 0081	9MO	M	BL	47,XY,+9p(9pter>9q11:)	U	A	PROBAND	602	1	Multiple malformations & mental retardation
CHROMOSOME 10													
*GM1429	8	MC5A	20U 0068	55	F	BL	47,X,+10,t(X;9)(Xpter>Xq12::9q33>9qter;9pter>9q33::Xq12>Xqter)	U	A	MOTHER	140	2	Mosaic; 16% of fibroblasts do not have additional 10
CHROMOSOME 13													
*GM0085	6	MC5A	20U 0143	2WK	M	BL	46,XY,-13,+t(13;13)(13qter>cen>13qter)	U	A	PROBAND			Clinical Trisomy 13
GM0503	2	MC5A	20U 0066	1	M		46,XY/47,XY,+13	U	A	PROBAND			54%/46%
GM0526	3	MC5A	20U 0095	NB	M	WH	47,XY,+13	U	A	PROBAND			
GM2948A	4	HF12	20U 0107	NB	M	WH	47,XY,+13	U	A	PROBAND			Microcephaly, cebocephaly, microophthalmia, & polydactyly
GM3084	9	EEMEM	20U 0224	11MO	M	BL	47,XY,+13	U	A	PROBAND			Multiple congenital anomalies
GM3330	12	EEMEM	20U 0044	1DA	M	WH	47,XY,+13	U	A	PROBAND			Multiple congenital anomalies
CHROMOSOME 14													
GM3233	2	HF12	20U 0122	1WK	M	WH	46,XY,r(14)/45,XY,-14	U	A	PROBAND			Congenital malformations; 88%/12%
CHROMOSOME 15													
GM3133	6	HF12	20U 0273	FW	F		46,X,+15	U	A	PROBAND			
GM3184	5	MC5A	20U 0273	11FW	M		47,XY,+15	U	A	PROBAND			
CHROMOSOME 18													
*GM0143	3	MC5A	20U 0107	3MO	F	BL	47,XX,+18	U	A	PROBAND			
GM0734	3	MC5A	20U 0095	18FW	F		47,XX,+18	U	A	PROBAND			
GM1118	8	EEMEM	20U 0150	30MO	F	WH	46,XX,r(18)/45,XX,-18	U	A	PROBAND			78% of cells have r(18)

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							ANEUPLOID						
CHROMOSOME 18													
GM1359	3	MC5A	20U 0107	4WK	M	WH	47,XY,+18	U	A	PROBAND			
GM2732	7	EEMEM	20U 0109	4DA	M	WH	47,XY,+18	U	A	PROBAND			Classical phenotype
GM3538	2	HF12	20U 0107	NB	F	WH	47,XX,+18	U	B	PROBAND			Classical syndrome findings
GM3623	6	MC5A	20U 0340	NB	F	BL	48,XXX,+18	U	A	PROBAND			
CHROMOSOME 21													
*GM0137	4	MC5A	20U 0075	6	M	WH	45,XY,-21	U	A	PROBAND	335	1	60% of normal superoxide dismutase-1; in some cells chromosome 4 shows extra, unidentifiable material which may be from chromosome 21
*GM0201	10	MC5A	20U 0030	13	F	WH	47,XX,+21,t(1;17)(1pter>1p32::17p13>17qter;17pter>17p13::1p32>1qter)	U	A	PROBAND	337	1	
*GM0230	9	EEMEM	10U 0088	14MO	F	WH	45,XX,-21	U	A	PROBAND			60% of normal superoxide dismutase-1
*GM0260	6	MC5A	20U 0095	11	M	WH	46,XY/47,XY,+21/47,XY,+t(21;21)(qter>cen>qter)	U	A	PROBAND	327	1	66%/24%/10% Mosaic; formerly GM0069
*GM1137	6	R1640	10U 0017	1MO	M	BL	48,XXY,+21,t(6;10)(6qter>6p22 or 24::10p12>10pter;10qter>10p12::6p22 or 24>6pter)	U	B	PROBAND			Down's syndrome features
GM2058	5	HF12	20U 0165	11DA	F	WH	46,XX/46,XX,-21,+t(21;21)(21qter>cen>21qter)	U	A	PROBAND			87%/13% at passage 8
GM2067	3	MC5A	10U 0095	20FW	M	WH	47,XY,+21	U	A	PROBAND			Down's syndrome phenotype
*GM2504	9	MC5A	20U 0095	1MO	M	BL	47,XY,+21	U	A	PROBAND	329	1	Formerly GM0258; also available at passage 5 (GM2504A); shows increased sensitivity to interferon
GM2524	7	MC5A	20U 0223	10WK	M	WH	47,XY,+21/46,XY,-21,+dic(21;21)(pter>q21::p12>cen>qter)	U	A	PROBAND			Mosaic; 82%/18%
GM2571	4	HF12	20U 0148	3MO	F	WH	48,XX,+21,+mar	U	A	PROBAND			Down's syndrome phenotype
GM2767	3	HF12	20U 0107	14	F		47,XX,+21	U	A	PROBAND			

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							ANEUPLOID						
CHROMOSOME 21													
GM3576	10	MC5A	20U 0326	13FW	M	WH	48,XY,+2,+21	U	A	PROBAND			
*GM1920	2	HF12	20U 0061	23	M	WH	47,XY,+21,inv(9)(pter>p13::q13> p13::q13>qter)mat	U	A	PROBAND	418	1	See GM1921 Lymphoid
CHROMOSOME 22													
*GM0084	5	MC5A	20U 0095	1MO	M	WH	47,XY,+22	U	A	PROBAND			Multiple congenital anomalies
							TERATOMA CELL LINES						
CHROMOSOME 00													
*GM1304		EEMEM	20U 0056	27	F		46,XX	B	A	PROBAND	154	1	Passage 4 at IMR; PGM1=1;PGM3=1;PGD=AB; chromosome polymorphisms: 3=+/+;13=+/+; 15=+/+;21=+/+; right ovarian teratoma
*GM1305		EEMEM	20U 0056	27	F		46,XX	B	B	SAME	154	1	Passage 4 at IMR;PGM1=1;PGM3=1;PGD=AB; chromosome polymorphisms: 3=+/-;13=+/-; 15=+/-;21=+/-; uterine tissue
*GM1306A		EEMEM	20U 0056	31	F	WH	46,XX	B	A	PROBAND	155	1	Passage 8 at IMR; PGM1=1;PGM3=2-1;PGD=A; chromosome polymorphisms:13=-/-;14=+/+; 21+-/-; right ovarian teratoma
*GM1307		EEMEM	20U 0056	31	F	WH	46,XX	B	B	SAME	155	1	Passage 5 at IMR; PGM1=1;PGM3=2-1;PGD=A; chromosome polymorphisms: 13=+/-;14=+/-; 21=+/-; fallopian tube tissue

SV40 VIRUS TRANSFORMED CELL CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
Cystinosis: Early Onset (Nephropathic Type; Infantile Type) - 21980												
*GM2894	47	HF12 10U	0114	9	F		--	B	PROBAND			SV40 transformed culture, aneuploid; accumulates large amounts of intracellular nonprotein cystine
Galactosemia (Transferase Deficiency) - 23040												
*GM0639	46	EEMEM 20U	0089	8	M	BL	--	A	PROBAND	26	1	SV40 transformed GM0052; Gal-1-P uridyl transf. absent after crisis, T-antigen positive; polyploid
*GM0638	48	EEMEM 20U	0089	3MO	M		--	A	PROBAND	27	1	SV40 transformed GM0054; Gal-1-P uridyl transf. absent after crisis, G6PD-B, T-antigen positive; hypodiploid
IMR90												
AG2804A	41	MC5A 20U	0135	16FW	F	WH			PROBAND			SV40 transformed IMR90; post-crisis, T-antigen positive, 1-5% V-antigen positive
Apparently Normal: Non-Fetal Tissue												
*GM0637	40	EEMEM 20U	0089	18	F	WH			PROBAND	162	2	SV40 transformed GM0037(Puerto Rican); T-antigen positive; polypoid
Retinoblastoma; Sporadic												
AG3022	49	HF12 20U	0135	27MO	F	WH			PROBAND			SV40 transformed AG1142; 100% T-antigen pos.,1-5% V-antigen pos.,post-crisis line near tetraploid;mult.rearrangement; deleted chrom. 13 present in 90% cells

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APPARENTLY NORMAL HUMAN LYMPHOCYTE CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
GM3299		R1640 20I	0309	8	F		PROBAND			
GM3714		R1640 15I	0343	10	F	WH	PROBAND	391	1	Sister of GM3715
*GM0892		R1640 20I	0024	12	F	WH	PROBAND			HLA type A2(A9),B12,B18
GM3715		R1640 15I	0343	12	F	WH	SIS	391	2	Sister of GM3714
GM3469		R1640 20I	0120	20	M	WH	FATHER	239	1	Mexican-American; 46,XY; daughter is GM3470 Lymphoid
GM0946		R1640 20I	0024	22	F	WH	PROBAND			46,XX
GM1953		R1640 20I	0089	22	F	WH	PROBAND	360	1	Formerly GM1079
GM2254		R1640 20I	0155	22	F	WH	PROBAND			46,XX
*GM0131		R1640 20I	0010	23	F	WH	PROBAND			46,XX
GM0333		R1640 20I	0010	23	F	WH	PROBAND			46,XX
*GM0546		R1640 20I	0089	23	F	WH	PROBAND			Spontaneously transformed; 46,XX
*GM0607		R1640 20I	0025	23	F	WH	PROBAND			46,XX,19p+
GM0922		R1640 20I	0024	23	F	WH	PROBAND	596	1	46,XX; same patient as GM0924
*GM0605		R1640 20I	0025	24	F	WH	PROBAND			47,XX,+15,del(1)(1pter>1p32::1p22>1qter); 32% of cells are missing an X chromosome
GM0923		R1640 20I	0024	24	M	WH	PROBAND			46,XY
*GM0130		R1640 20I	0010	25	M	WH	PROBAND			46,XY
*GM0558		R1640 20I	0089	26	M	WH	PROBAND			HLA type A3,A11,Bw15,Bw22
GM1805		R1640 20I	0089	26	F	WH	PROBAND	355	1	HLA type A1,A9,B18(B13); formerly GM1073
GM3201		R1640 20I	0208	26	M	WH	PROBAND			Clinically normal; absent neutral alpha-glucosidase C activity
GM0536		R1640 20I	0089	27	M	WH	PROBAND			
*GM0621		R1640 20I	0089	28	M	WH	PROBAND			
GM1078		R1640 20I	0089	30	F	WH	PROBAND			
GM0893		R1640 20I	0024	32	F	WH	PROBAND	595	1	46,XX; same patient as GM0894

APPARENTLY NORMAL HUMAN LYMPHOCYTE CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
GM1806		R1640 20I	0089	33	M	WH	PROBAND	356	1	Formerly GM1074; HLA type A1,A2,Bw17, B18,(Bw22)
*GM1989		R1640 20U	0089	33	M	WH	PROBAND	358	1	Formerly GM1076
*GM2131		R1640 20I	0205	33	M	WH	PROBAND			ATCC CCL 156; 45,XY(-G); secretes IgM-Lambda chains, HLA type A2,A7
GM2184		R1640 20I	0186	36	M	WH	PROBAND	455	1	Normal spouse of HD patient; see GM2185 Fibroblast
*GM1814		R1640 20I	0089	37	F	WH	PROBAND	354	1	Formerly GM1072; HLA type (A1),A2,A11, B8,Bw40
GM1815		R1640 20I	0089	42	M	WH	PROBAND	357	1	Formerly GM1075; HLA type A2,A9,B7,B13, (B12)
GM1954		R1640 20I	0089	44	F	WH	PROBAND	361	1	Formerly GM1080
*GM1310		R1640 20I	0089	51	M	WH	PROBAND			
*GM1990		R1640 20I	0089	51	F	WH	PROBAND	359	1	Formerly GM1077
*GM1056		R1640 20I	0026	65	M	WH	PROBAND			HLA type A9,29,B12,Bw15
GM3657		R1640 20I	0026	68	M	WH	PROBAND	542	1	See GM3658 Fibroblast

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Argininemia - 20780												
*GM2011		R1640 20I	0006	5	F	WH	--		PROBAND	422	1	Mexican-American; see also GM0954 Fibroblast; RBC arginase deficiency
Argininosuccinicaciduria - 20790												
GM3010		R1640 20I	0207	8	F		--		PROBAND			Absent RBC argininosuccinic acid lyase
Carnosinemia - 21220												
GM2631		R1640 20I	0008	12	M	WH	--		PROBAND	363	1	46,XY; 30% of normal plasma carnosinase activity
Citrullinuria (Citrullinemia) - 21570												
*GM0235		R1640 20I	0010	31	M	WH	--	B	PROBAND			HLA type (Aw31,33)A9,Bw15,B12; deficient argininosuccinate synthetase; 46,XY; line UM-21
GM1204		R1640 20I	0107	8MO	M	WH	--		PROBAND	2	1	See GM1044 Fibroblast (biochemically verified); HLA type A2,B12/A28,Bw35
GM1206		R1640 20I	0107	27	M	WH	+-(0)		FATHER	2	2	See GM1058 Fibroblast; HLA type A11,Bw15/A2,B12
GM1205		R1640 20I	0107	26	F	WH	+-(0)		MOTHER	2	3	HLA type A28,Bw35/A1,B13
GM1207		R1640 20I	0107	5	F	WH	+?		SIS	2	5	HLA type A11,Bw15/A28,Bw35
GM1685		R1640 20I	0157	1DA	M	WH	--	A	PROBAND	3	1	See GM1679 Fibroblast; HLA type A1,Aw32,B18,B27; deficient argininosuccinate synthetase; a sib died of citrullinuria
Cystathioninuria - 21950												
GM1454		R1640 20I	0008	7		OR	--		PROBAND			Unresponsive to B6; HLA type A28,A2,Bw40,Bw21
GM1562		R1640 20I	0008	42		WH	+-(0)		PARENT			HLA type A3,A10,B13,B18
GM1781		R1640 20I	0008	18		WH	--		PROBAND	169	1	B6 responsive
GM1461		R1640 20I	0008	39		WH	+-(0)		PARENT	169	2	HLA type A2,A3,Bw22,Bw40
GM1565		R1640 20I	0008	16	F	WH	--		PROBAND	170	1	Also has PKU; HLA type A28,Aw24,B7,Bw35
GM1807		R1640 20I	0008		M	WH	+-(0)		FATHER	170	2	HLA type A1,A2,Bw35,B13
GM1566		R1640 20I	0008	16		WH	--		PROBAND	406	1	HLA type A1,A28,B13,Bw35
GM1456		R1640 20I	0008	43		WH	+-(0)		PARENT	406	2	HLA type A1,B13,Bw27
GM1868		R1640 20I	0008	49		WH	+-(0)		PARENT	406	3	HLA type A28,A10,Bw15,Bw16

HUMAN LYMPHOCYTE CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM											
Gyrate Atrophy of Choroid and Retina (Ornithinemia) - 25887											
*GM3534		R1640 20I	0334	37	F	WH	--	PROBAND			Portuguese; deficient ornithine aminotransferase activity in PHA stimulated lymphocytes and fibroblasts
GM3540		R1640 20I	0334	36	F	WH	--	PROBAND			Scotch-Irish-English; deficient ornithine aminotransferase activity in fibroblasts
Homocystinuria (Cystathionine Synthetase Deficiency) - 23620											
GM1446		R1640 20I	0008		F	WH	+-(0)	PARENT			HLA type A2,3,Bw35,Bw17
GM1447		R1640 20I	0008		F	WH	+-(0)	PARENT			HLA type A1,A10,B7,(Bw16)
GM1463		R1640 20I	0008	42	F	WH	+-(0)	PARENT			HLA type A2,A11,B27,Bw22
GM1528		R1640 20I	0008		M		+-(0)	PARENT			HLA type A10,(A3),B18,Bw22
GM1529		R1640 20I	0008	43	F		+-(0)	PARENT			HLA type A10,Aw23,B8,B18
GM1532		R1640 20I	0008		M		--	PROBAND			HLA type A2,B7,Bw22,(B18)
GM1558		R1640 20I	0008		F		+-(0)	PARENT			HLA type A10,Aw24,B7,Bw15
GM1559		R1640 20I	0008		M		+-(0)	PARENT			HLA type A3,A10,Bw15,B27
GM1560		R1640 20I	0008		M		--	PROBAND			HLA type A2,3,B7,B18
GM1808		R1640 20I	0008	45	M	WH	+-(0)	PARENT			HLA type A2,A10,B12,B27
GM2689		R1640 20I	0038	25	M	WH	--	PROBAND	241	1	
GM2688		R1640 20I	0038	27	M	WH	+?	BRO	241	2	Clinically normal; possible heterozygote
GM2690		R1640 20I	0038		M	WH	+-(0)	FATHER	241	3	
GM2429		R1640 20I	0008		F	WH	--	PROBAND	480	1	Formerly GM2375
Maple Syrup Urine Disease - (Branched-Chain Ketoaciduria) - 24860											
GM1366		R1640 20I	0107	7	F	BL	--	PROBAND	20	1	See GM1364 Fibroblast; HLA type A2(A9),B8,B12; classical symptoms; one previously affected male sib
GM1655		R1640 20I	0107	5	F	WH	--	PROBAND	21	1	See GM1654 Fibroblast; HLA type A1,A2,B13,Bw17(B23)(Bw40); Mennonite kindred; two female sibs died of MSUD
Phenylketonuria - 26160											
GM2659		R1640 20I	0038	14	F	WH	--	PROBAND			Puerto Rican

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Amyloidosis - Dominant - 10520												
GM1999		R1640 20I	0155	16	F	WH	+-		PROBAND	421	1	Non-Portuguese; see GM1998 Fibroblast
Disaccharide Intolerance I (congenital sucrose-isomaltose malabs) - 22290												
GM3070		R1640 20I	0297	14	M	WH	--		PROBAND			
Fucosidosis - 23000												
GM1025		R1640 20I	0008	4	M	WH	--	B	PROBAND	24	1	See GM0292 Fibroblast; deficient Alpha-L-Fucosidase; Type 2
GM1024		R1640 20I	0008	9	M	WH	--	B	AFF BRO	24	2	Type 2; see GM0291 Fibroblast; deficient Alpha-L-Fucosidase
GM1026		R1640 20I	0008	29	F	WH	+-(O)	B	MOTHER	24	3	See GM0290 Fibroblast; low-normal Alpha-L-Fucosidase activity
GM1023		R1640 20I	0008	31	M	WH	+-(O)	B	FATHER	24	4	See GM0289 Fibroblast; low-normal Alpha-L-Fucosidase activity
Galactosemia (Transferase Deficiency) - 23040												
GM2796		R1640 20I	0260	15	M	BL	--		PROBAND	26	1	See GM0052, GM1908 Fibroblast & GM0639 (SV40 Transformed)
GM2795		R1640 20I	0095	7	M	WH	--		PROBAND	27	1	See also GM0054, GM1907 Fibroblast and GM0638 (SV40 Transformed)
GM2412		R1640 20I	0008	15	M	WH	--	B	PROBAND	28	1	Formerly GM1027; HLA type A2,B12/A1,B8; deficient galactose-1-phosphate uridyl transferase;see GM0441 Fibroblast
GM1028		R1640 20I	0008	16	F	WH	--		AFF SIS	28	2	HLA type A1/A2,B12;see GM0440 Fibroblast
GM1029		R1640 20I	0008	41	F	WH	+-(O)		MOTHER	28	3	HLA type A2,B12/A28,Bw35; see GM0439 Fibroblast; 50% of normal transferase activity in erythrocytes
GM0148		R1640 20I	0010	8	M	WH	--	B	PROBAND	171	1	46,XY; deficient galactose-1-phosphate uridyl transferase
GM0149		R1640 20I	0010	47	M	WH	+-	B	FATHER	171	2	46,XY; reduced galactose-1-phosphate uridyl transferase
Glycogen Storage Disease: Type II - Pompe Disease - 23230												
GM1464		R1640 20I	0008	46	F	WH	--	B	PROBAND	173	1	Acid maltase deficiency; adult form
GM1778		R1640 20I	0008	16	F	WH	+-(O)		DAU	173	2	
GM1568		R1640 20I	0008	71	M	WH	+-	B	FATHER	173	4	Heterozygote enzyme level

HUMAN LYMPHOCYTE CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Glycogen Storage Disease: Type VII -Muscle PFK Deficiency - 23280												
GM3128		R1640 20I	0317	31	M	WH	--		PROBAND			Muscle deficient in phosphofructokinase (M4)
Mucolipidosis: Type II - I-Cell Disease - 25250												
GM2660		R1640 20I	0008	23	F		+-(O)		PARENT	469	3	Child has I-cell disease
Mucolipidosis: Type IV - 25265												
*GM2533		R1640 20I	0077	2	F	WH	--	B	PROBAND	229	1	Ashkenazi; see GM2048 Fibroblast; affected sib is GM2529 Amniotic
Mucopolysaccharidosis: Type IH - Hurler Syndrome - 25280												
GM1034		R1640 20I	0008	2	F	WH	--		PROBAND	599	1	HLA type A2,Aw30,B12,Bw22; same patient as GM1867
Mucopolysaccharidosis: Type IH/S - Hurler/Scheie - 25280												
GM1032		R1640 20I	0008	15	M	IN	--		PROBAND			HLA type A9,A10,B13
Mucopolysaccharidosis: Type IIIA - Sanfilippo Syndrome, A - 25290												
GM1780		R1640 20I	0008	8	M	WH	--		PROBAND			
Mucopolysaccharidosis: Type VI - Maroteaux-Lamy Syndrome - 25320												
*GM1022		R1640 20I	0008	4	F	WH	--	B	PROBAND	51	1	Deficient arylsulfatase B; see GM0519 Fibroblast
GM3721		R1640 20I	0016	25	F	BL	+-(O)		MOTHER	392	1	See GM3720 Fibroblast
GM3723		R1640 20I	0016	40MO	F	BL	--		PROBAND	392	2	5% of normal arylsulfatase B activity; see GM3722 Fibroblast
GM3725		R1640 20I	0016	41	M	BL	+-(O)		FATHER	392	3	See GM3724 Fibroblast
GM3727		R1640 20I	0016	9	F	BL	+?		SIS	392	4	Possible heterozygote; see GM3726 Fibroblast
DISORDERS OF LIPID METABOLISM												
Abetalipoproteinemia - 20010												
*GM1453		R1640 20I	0008	18	F	WH	--		PROBAND			
*GM1810		R1640 20I	0008	23	M	WH	--		PROBAND	398	1	HLA type A1,A10,B7,B13; formerly GM1451
GM2 Gangliosidosis: Tay-Sachs Disease, AB Variant - 27275												
GM3691		R1640 15I	0328	13	F	WH	--		PROBAND			Dysarthria, dementia, & ataxia; normal B-hexosaminidase A & B, B-galactosidase, arylsulfatase A, & glycoprotein neuraminidase activities

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM												
GM2 Gangliosidosis: Type I - Tay-Sachs Disease - 27277, 27280												
GM2366		R1640 20I	0008	28	M	WH	+-	B	PROBAND			Carrier; heterozygous levels of hexosaminidase A
GM3441		R1640 20I	0328	37	F	WH	--?		PROBAND			Variant; hexosaminidase A deficiency; dementia; ataxia; upper & lower motor neuron involvement; Ashkenazi
GM3461		R1640 20I	0032	24	M	WH	--?		PROBAND			Ashkenazi; variant; deficient hexosaminidase A; weakness and ataxia
GM3575		R1640 20I	0328	31	F	WH	--?		PROBAND			Ashkenazi; ataxia, dementia, peripheral neuropathy; deficient hexosaminidase A; variant
GM3585		R1640 20I	0032	13MO	F	WH	--	B	PROBAND			5% of normal hexosaminidase A activity
GM3586		R1640 20I	0032	36MO	F	WH	--	B	PROBAND			8% of normal hexosaminidase A activity
GM3770		R1640 20I	0328	15	M	WH	--?		PROBAND	494	1	Jewish; variant; hexosaminidase A deficiency in white cells; mild dysarthria
GM3771		R1640 20I	0328	13	M	WH	--?		AFF BRO	494	2	Jewish; severe tremor, moderate ataxia, mild dysarthria; hexosaminidase A deficiency in white cells
GM3772		R1640 20I	0328	11	M	WH	--?		AFF BRO	494	3	Jewish; mild ataxia and hand tremors; hexosaminidase A deficiency in white cells; twin of GM3773
GM3773		R1640 20I	0328	11	M	WH	--?		AFF BRO	494	4	Jewish; mild ataxia and hand tremors; hexosaminidase A deficiency in white cells; twin of GM3772
Gaucher Disease: Type 3 (Juvenile and Adult, Cerebral) - 23100												
GM1019		R1640 20I	0008	35	M	WH	+-	B	PARENT			Father of a Juvenile Gaucher; low-normal acid B-glucosidase
GM1769A		R1640 20I	0008	25	M	WH	--	B	PROBAND	249	1	Adult Gaucher; formerly GM1021; low acid B-glucosidase
GM1020		R1640 20I	0008	49	F	WH	+-	B	MOTHER	249	2	Low-normal acid B-glucosidase
GM1030		R1640 20I	0008	50	F	WH	+-	B	PARENT	250	1	Mother of a Juvenile Gaucher; low-normal acid B-glucosidase
GM1031		R1640 20I	0008	53	M	WH	+-	B	PARENT	250	2	Father of a Juvenile Gaucher; low-normal acid B-glucosidase

HUMAN LYMPHOCYTE CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
					DISORDERS OF LIPID METABOLISM							
Combined Hyperlipidemia - 14425												
*GM1455		R1640 20I	0008	33	M		+-		PROBAND			HLA type A11(A3),Bw22,(Bw40)
GM1783		R1640 20I	0008	31	F		+-		PROBAND	296	1	Formerly GM1457
Familial Hypercholesterolemia - 14389 & Hyperlipoproteinemia: Type II-14440												
*GM1458		R1640 20I	0008	30	F		--		PROBAND			Receptor negative by clinical diagnosis
GM1460		R1640 20I	0008		F		+-		PARENT			Mother of a clinically diagnosed receptor negative child
GM1567		R1640 20I	0008	28	M	BL	--		PROBAND			Abnormal regulation of 3-hydroxy-3-methylglutaryl coenzyme A reductase and incorporation of oleate into cholesteryl esters
*GM1767		R1640 20I	0008	19	F		--	B	PROBAND	264	1	Formerly GM1452; receptor negative
GM1784		R1640 20I	0008	23	M		--		PROBAND	265	1	Formerly GM1462
GM2373		R1640 20I	0008	42	M	WH	+-		PARENT	271	7	
GM1459		R1640 20I	0008	25	F	WH	--	A	PROBAND	387	1	See GM0486 Fibroblast; lymphoid line is verified receptor negative
*GM1448		R1640 20I	0008	18	F	WH	+-		PROBAND	506	1	Elevated serum cholesterol
GM2110		R1640 20I	0008	49	M	WH	+-		AFF FATHER	506	2	Formerly GM1450; tendon xanthoma; premature myocardial infarctions; elevated cholesterol levels (above 400 mg%); died before age 50
GM1766		R1640 20I	0008	23	F	WH	+-		AFF SIS	506	3	Formerly GM1449; elevated serum cholesterol
Metachromatic Leukodystrophy: Infantile - 25010												
GM1018		R1640 20I	0008	25	F	WH	+-	B	PARENT			Mother of infantile MLD; reduced activity of arylsulfatase A
Metachromatic Leukodystrophy: Juvenile - 25020												
GM1017		R1640 20I	0008	14	F	WH	--	B	PROBAND	622	1	HLA type A2,A29,B12,B16; same patient as GM1785; deficient Arylsulfatase A
GM1016		R1640 20I	0008	42	M	WH	+-(O)		FATHER	622	2	
Multiple Sulfatase Deficiency Disease - 27220												
*GM3246		R1640 20I	0105	9	M	WH	--	B	PROBAND	632	1	See GM3245 Fibroblast; deficient Arylsulfatase A, B, and C

HUMAN LYMPHOCYTE CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM											
Niemann-Pick Disease: Type C - 25720											
GM3124		R1640 20I	0107	9	F	WH	--	PROBAND	451	1	See GM3123 Fibroblast; 38% of normal sphingomyelinase activity in fibroblasts
Sea-Blue Histiocyte Disease - 26960											
GM1913		R1640 20I	0348	24	F	WH	--	PROBAND	407	1	See GM1912 Fibroblast; HLA type A2,Aw31, B18,B13; muscle biopsy showed foamy histiocytes & sea-blue histiocytes; eye movement abnormalities
DISORDERS OF METAL METABOLISM											
Menkes Syndrome (Kinky Hair Disease) - 30940											
GM1245		R1640 20I	0107	5DA	M	WH	y-	AFF BRO	79	2	See GM1057 Fibroblast; HLA type A9,A10, B13,B18; fibroblasts show increased copper concentration
GM1982		R1640 20I	0107	2	M	WH	y-	PROBAND	80	1	See GM1981 Fibroblast; fibroblasts show elevated copper concentration
GM1984		R1640 20I	0107	26	F	WH	+?	MOTHER	80	2	See GM1983 Fibroblast; son is only affected child in an extended family
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM											
Adenosine Deaminase Deficiency, with Immunodeficiency - 24275											
*GM2756		R1640 20I	0236	3	M	BL	--	A PROBAND			Deficient ADA activity
*GM2471		R1640 20I	0219		M		--	A PROBAND	223	1	Deficient ADA activity
GM2472		R1640 20I	0219		F		+-	A MOTHER	223	2	Heterozygote level of ADA activity
*GM2606		R1640 20I	0223	8MO	M	WH	--	A PROBAND	226	1	See GM2605 Fibroblast; deficient ADA activity
GM2608		R1640 20I	0223		F	WH	+-	A MOTHER	226	2	See GM2607 Fibroblast
GM2610		R1640 20I	0223		M	WH	+-	A FATHER	226	3	See GM2609 Fibroblast
GM2581		R1640 20I	0223	38	M	WH	+-	A FATHER	232	2	See GM2580 Fibroblast
GM2583		R1640 20I	0223	37	F	WH	+-	A MOTHER	232	3	See GM2582 Fibroblast
*GM1715		R1640 20I	0217	33MO	M	BL	--	A PROBAND	423	1	Detroit 300; about 2% of normal ADA activity; HLA type:A2,9;Bw40,8; see GM2445 Fibroblast
*GM2825A		R1640 20I	0291	4MO	F	WH	--	B PROBAND	446	1	Absent IgM and IgA; weak MLC response; absent T cells; see GM2824 Fibroblast; no detectable ADA activity in erythrocytes

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DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Adenosine Deaminase Deficiency, with Immunodeficiency - 24275												
*GM2294		R1640 20I	0208	10MO	M	BL	--	A	PROBAND	633	1	See GM1997 Fibroblast; partial ADA deficiency
Adenosine Deaminase Deficiency, with no Immunodeficiency - 24276												
*GM3043		R1640 20I	0170	16	M	BL	--		PROBAND	634	1	Kung South African; see GM3029 Fibroblast; clinically normal
Inosine Triphosphate Pyrophosphohydrolase (ITPase) Deficiency - 14752												
GM1619		R1640 20I	0177	29	F	WH	+-		PROBAND	635	1	See GM1617 Fibroblast; deficient RBC ITPase
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800												
*GM0467		R1640 20I	0083		F				MUTANT			Chemically induced in vitro with E.M.S.; selectable with 8-azaguanine or 6-thioguanine
*GM2292		R1640 20I	0183	19FW	M	WH	y-		PROBAND	74	1	See GM2290 & GM2291 Fibroblast; also GM2338 Amniotic
GM1899		R1640 20I	0066	10	M	WH	y-		PROBAND	623	1	HLA type A2,A11,B18,B5(Bw35)
GM1900		R1640 20I	0066		F	WH	+?		MOTHER	623	2	HLA type A2,A28,B18,B5(Bw35)
Xeroderma Pigmentosum: Complementation Group A - 27870												
*GM2345		R1640 20I	0211	8	F	OR	--		PROBAND			Japanese; XP20J
*GM2250		R1640 20I	0155	10	F	WH	--		PROBAND	484	1	XP12BE; same patient as GM2500; less than 2% of normal UV induced unscheduled DNA synthesis in fibroblasts
Xeroderma Pigmentosum: Complementation Group B - 27870												
*GM2252		R1640 20I	0155	31	F	WH	--		PROBAND	283	1	Formerly GM1853; XP11Be; also affected with Cockayne syndrome; 3-7% of normal UV induced unscheduled DNA synthesis in fibroblasts; HLA type Aw32,w24,Bw40,B12
*GM1855		R1640 20I	0155	64	F	WH	+-(O)		MOTHER	283	2	See GM1854 Fibroblast; also heterozygous for Cockayne Syndrome; HLA type Aw32,(A1),B12,B27
Xeroderma Pigmentosum: Complementation Group C - 27870												
*GM2249		R1640 20I	0155	16	M	WH	--		PROBAND	75	1	XP8BE; 46,XY; 10-20% of normal UV induced unscheduled DNA synthesis in fibroblasts; see GM0671 Fibroblast
*GM2498		R1640 20I	0155	16	M	WH	--		AFF TWIN	75	2	XP9BE; 46,XY; 10-20% of normal UV induced unscheduled DNA synthesis in fibroblasts; see GM0676 Fibroblast

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DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM											
Xeroderma Pigmentosum: Complementation Group C - 27870											
*GM2246		R1640 20I	0155	30	F	WH	--	PROBAND	482	1	XP1BE; 46,XX; 10-20% of normal UV induced unscheduled DNA synthesis in fibroblasts; same patient as GM2483
*GM2248		R1640 20I	0155	25	M	WH	--	PROBAND	507	1	XP3BE; 46,XY; 10-20% of normal UV induced unscheduled DNA synthesis in fibroblasts; see GM0030 Fibroblast
GM2634		R1640 20I	0226	16	F	BL	--	PROBAND	627	1	DeSanctis-Cacchione; Lupus erythematosus; XP1MI; see GM2096 Fibroblast
Xeroderma Pigmentosum: Complementation Group D - 27870											
*GM2485		R1640 20I	0155	14	F	WH	--	PROBAND	624	1	XP7BE; 25-50% of normal UV induced unscheduled DNA synthesis in fibroblasts; same patient as GM2590; 46,XX
*GM2253		R1640 20I	0155	14	M	BL	--	PROBAND	625	1	XP17BE; 25% of normal UV induced unscheduled DNA synthesis in fibroblasts; 46,XY
GM2473		R1640 20I	0155	34	F	BL	+-(0)	MOTHER	625	2	
Xeroderma Pigmentosum: Complementation Group E - 27870											
*GM2450		R1640 20I	0155	30	F	WH	--	RELATIVE	626	2	XP3RO; relative of XP2RO (GM2415 Fibroblast); 46,XX
Xeroderma Pigmentosum: Complementation Group Undetermined - 27870											
GM2449		R1640 20I	0110	30	M	WH	--	PROBAND			XPPHBE; variant type; 46,XY
GM2344		R1640 20I	0173	42MO	M	WH	--	PROBAND	77	1	XP1WI; see GM1630 Fibroblast
GM1646		R1640 20I	0133	21	F	WH	--	PROBAND	377	1	See GM1389 Fibroblast
*GM2545		R1640 20I	0228	15	M	OR	--	PROBAND	499	1	Hawaiian
Pigmented Xerodermoid											
*GM3456		R1640 20I	0323	86	F	WH	-?	PROBAND	220	1	X.P. variant; late onset; normal excision repair; prolonged post UV DNA synthesis; see GM3379 Fibroblast
DISORDERS OF STEROID METABOLISM											
Adrenal Hyperplasia (Adrenogenital Syndrome) Type III - 20191											
GM2242		R1640 20I	0107	9MO	F	WH	--	PROBAND	85	1	See GM2241 Fibroblast; 21-hydroxylase deficiency; salt-losing type; responsive to hydrocortisone treatment
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE											
Cockayne Syndrome - 21640											
*GM2502		R1640 20I	0155	13	F	WH	--	PROBAND			Slow growing culture; 46,XX

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						DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE						
Cockayne Syndrome - 21640												
*GM1712		R1640 20I	0006	21	M	WH	--		PROBAND	251	1	HLA type Aw32,A10,Bw15,B14,(A3); UV irradiation sensitive; see GM1098 Fibroblast
GM1857		R1640 20I	0155	13	M	WH	--		PROBAND	399	1	HLA type: A9,A11;B7,Bw15; see GM1856 Fibroblast
GM2474		R1640 20I	0155	29	M	WH	+-(O)		FATHER	628	2	Affected daughter is GM1629 Fibroblast
GM2964		R1640 20I	0210	25	F	WH	--		PROBAND	636	1	Atypical; late age of onset; lack of ocular and retinal lesions; see GM2965 Fibroblast
Dystonia Musculorum Deformans, Dominant Form - 12810												
GM2217		R1640 20I	0184	30	M	WH	+-		PROBAND	100	1	Age of onset 24 yrs; on Lithium and Valium treatment
GM2305		R1640 20I	0184	17	F	WH	+-		PROBAND	102	1	See GM2304 Fibroblast; age of onset 7 yrs; at time of venipuncture has very little speech and considerable dystonia of all extremities
GM2347		R1640 20I	0184	21	F	WH	+-		PROBAND	218	1	Swedish background; age of onset 5 yrs; see GM2348 Fibroblast
GM2264		R1640 20I	0184	14	F	WH	+-		PROBAND	475	1	Jewish background; age of onset 8 yr; minimal action dystonia; greatest difficulty with speech and chewing; parents are healthy
Dystonia Musculorum Deformans, Recessive Form - 22450												
GM2307		R1640 20I	0184	13	M	WH	--		PROBAND	103	1	See GM2306 Fibroblast; age of onset 10 yrs; Jewish background
GM2256		R1640 20I	0184	14	F		--		PROBAND	473	1	See GM2255 Fibroblast; age of onset 12 yrs; severe involvement of trunk and extremities; similarly affected sister
Muscular Dystrophy; Duchenne Type (Pseudohypertrophic Progressive) - 31020												
GM3780		R1640 20I	0006	11	M	WH	y-		PROBAND	533	1	Latin American; elevated serum CPK; see GM3781 Fibroblast
GM3782		R1640 20I	0006	10	M	WH	y-		AFF BRO	533	2	Latin American; elevated serum CPK; see GM3783 Fibroblast
Myotonic Dystrophy (Steinert Disease) - 16090												
GM3094		R1640 20I	0210	27	M	WH	+-		PROBAND			Skeletal muscle myotonia; mild muscle weakness; abnormal glucose tolerance test

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DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Myotonic Dystrophy (Steinert Disease) - 16090												
GM3756		R1640 20I	0348	44	M	WH	+-		PROBAND	488	1	Typical phenotype; mother, two brothers, and a sister also affected; see GM3755 Fibroblast
GM3696A		R1640 20I	0348	12	F	WH	+-		AFF DAU	527	2	Family history of clinical phenotype; EMG myotonia and mild myopathic facies; affected father is GM3759 Fibroblast
OTHER DISORDERS OF KNOWN BIOCHEMISTRY												
Porphyria: Hereditary Coproporphyria - 12130												
GM2302		R1640 20I	0112	59	F	WH	+-		PROBAND	641	1	See GM0962 Fibroblast
Porphyria: Acute Intermittent Porphyria - 17600												
*GM1363		R1640 20I	0112		F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase; active AIP
GM2124		R1640 20I	0112	32	F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase; active AIP
GM2213		R1640 20I	0112	62	F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase; active AIP
GM2229		R1640 20I	0112	23	F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase; latent carrier
GM2230		R1640 20I	0112	58	F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase; latent carrier
GM2135		R1640 20I	0112	34	F	WH	+-	A	PROBAND	82	1	See GM0939 Fibroblast; deficient uroporphyrinogen-I synthetase; active AIP
GM2134		R1640 20I	0112	63	F	WH	++	A	MOTHER	82	2	See GM0940 Fibroblast; clinically normal; no URO synthetase deficiency
GM2133		R1640 20I	0112	65	M	WH	+-	A	FATHER	82	3	See GM0941 Fibroblast; deficient URO synthetase; clinically normal; latent carrier
Porphyria: Variegata - 17620												
GM2303		R1640 20I	0112	59	F	WH	+-		PROBAND			
Thalassemias - 27350												
*GM2064		R1640 20I	0192	53	M	BL	--	A	PROBAND			Ghanian; homozygous for hereditary persistance of fetal Hb, heterozygous for elliptocytosis

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OTHER DISORDERS OF KNOWN BIOCHEMISTRY												
Thalassemias - 27350												
*GM2232		R1640 20I	0192	24	M	WH	+-		PROBAND	470	1	Greek; hereditary persistance of fetal hemoglobin and B-thalassemia trait
*GM2266		R1640 20I	0192	17	F	WH	--	A	PROBAND	514	1	Sicilian; homozygous delta-beta thalassemia
*GM2267		R1640 20I	0192	19	F	WH	--	A	AFF SIS	514	2	Sicilian; homozygous delta-beta thalassemia
*GM2568		R1640 20I	0192				--	A	PROBAND	520	1	Homozygous for hemoglobin Lepore
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Acute Lymphocytic Leukemia												
*GM2219A		R1640 10I	0260	19	M				PROBAND			Line Molt-4, T lymphoblastoid cell line
*GM3671A		R1640 20I	0260	3	F	WH			PROBAND			T lymphoblastoid cell line CCRF-CEM; free of virus particles, non-immunoglobulin producing; near diploid
*GM3638		R1640 20I	0341	16	F				PROBAND	195	1	B cell line (line 8392); same patient as GM3639
*GM3639		R1640 20I	0341	16	F				PROBAND	195	1	T cell line (line 8402); same patient as GM3638
Allergic Asthmatic												
GM0604		R1640 20I	0025	10	M	WH			PROBAND			
Antitrypsin Deficiency - 20740												
GM3580		R1640 20I	0190	34	M	WH	+-		FATHER	209	1	Pi type MZ in serum; Greek
GM3579		R1640 20I	0190	31	F	WH	+-		MOTHER	209	2	Pi type MZ in serum; Cuban
GM3578		R1640 20I	0190	3	F	WH	--		PROBAND	209	3	Pi type ZZ in serum; Cuban-Greek
Basal Cell Nevus Syndrome - 10940												
GM1656		R1640 20I	0104	53	M	WH	+-		PROBAND	88	1	See GM1657 & GM1658 Fibroblast
GM1726		R1640 20I	0104	58	F	WH	+-		PROBAND	89	1	See GM1725 Fibroblast; HLA type A28, Aw30,B5,Bw16
GM2099		R1640 20I	0104	31	M	WH	+-		PROBAND	90	1	See GM2098 Fibroblast
GM2139		R1640 20I	0104		F	WH	+-		PROBAND	91	1	See GM2138 Fibroblast
GM1578		R1640 20I	0104	39	M	WH	+-		PROBAND	372	1	See GM1577 Fibroblast

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Basal Cell Nevus Syndrome - 10940											
GM1576		R1640 20I	0104	31	F	WH	+-	PROBAND	373	1	See GM1575 Fibroblast
GM1553		R1640 20I	0104	27	M	WH	+-	PROBAND	637	1	See GM1552 Fibroblast; HLA type A28,A3, Bw22,B13(B12)
Carbonic Anhydrase, Erythrocyte; Electrophoretic Variant - 11480											
*GM2744		R1640 20I	0234	17	F	WH	--	PROBAND	629	1	Presumed homozygous for red cell carbonic anhydrase I deficiency; clinically normal; Greek
*GM2742		R1640 20I	0234	44	M	WH	+-(O)	FATHER	629	2	Greek
*GM2743		R1640 20I	0234	37	F	WH	+-(O)	MOTHER	629	3	Greek
*GM2745		R1640 20I	0234	16	M	WH	--	AFF BRO	629	4	Presumed homozygous for red cell carbonic anhydrase I deficiency; clinically normal; Greek
Chediak-Higashi Syndrome - 21450											
*GM3365		R1640 20I	0227	27	M	WH	--	PROBAND			Albinism; recurrent skin infections; neuropathy; mild mental retardation
GM2431		R1640 20I	0216	32	M	WH	--	PROBAND	630	1	Formerly GM2378
GM2430		R1640 20I	0216	55	F	WH	+-(O)	MOTHER	630	2	Italian
GM2432		R1640 20I	0216	28	F	WH	+?	SIS	630	3	Clinically normal; possible heterozygote
Coffin-Lowry Syndrome - 30360											
GM3317		R1640 20I	0107	8	M	WH	y-	PROBAND	510	1	See GM3318 Fibroblast; mental retardation; coarse facies; positive family history
Cystic Fibrosis (Mucoviscidosis) - 21970											
GM0504		R1640 20I	0025	19	M	WH	--	PROBAND			Cell free supernatant produces ciliary dyskinesis factor in rabbit bioassay; metachromasia positive
GM0505		R1640 20I	0025	37	F	WH	+-(O)	PARENT			Cell free supernatant produces ciliary dyskinesis factor in rabbit bioassay; metachromasia positive
GM0506		R1640 20I	0025	8	F	WH	--	PROBAND			Cell free supernatant produces ciliary dyskinesis factor in rabbit bioassay; metachromasia negative
GM0608		R1640 20I	0025	21	M	WH	--	PROBAND			Metachromasia positive

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
GM1444		R1640 20I	0008	32	F		+-(O)	PARENT			Culture produces ciliary dyskinesis factor
GM1530		R1640 20I	0008	5	M		--	PROBAND			HLA type A2,A11,Bw17,Bw35; metachromasia positive
GM1445		R1640 20I	0008	3	F	WH	--	PROBAND	175	1	Culture produces ciliary dyskinesis factor; metachromasia negative; one similarly affected sib; Jewish extraction
GM1443		R1640 20I	0008	28	F	WH	+-(O)	MOTHER	175	2	Culture produces ciliary dsykinesis factor; metachromasia negative; Polish-Russian extraction
GM1442		R1640 20I	0008	31	M	WH	+-(O)	FATHER	175	3	Culture produces ciliary dyskinesis factor; metachromasia negative; Hungarian-Polish-Russian extraction
GM0897		R1640 20I	0089	13	M	WH	--	PROBAND	404	1	See GM0768 Fibroblast; mild; no pancreatic involvement
GM1531		R1640 20I	0008	23	M	WH	--	PROBAND	426	1	HLA type A2,(A9),B7,(Bw40); see GM0851 Fibroblast
Diabetes Mellitus of the Pima Indian - 22210											
GM2575		R1640 20I	0215	23	M	AI		PROBAND	230	1	Pima-Papago American Indian; onset at age 18; on Insulin; see GM2574 Fibroblast
GM2566		R1640 20I	0215	28	M	AI		PROBAND	231	1	Onset at age 17 yrs; not on Insulin; see GM2565 Fibroblast
GM2417		R1640 20I	0215	42	F	AI		PROBAND	383	1	Mother & siblings are also affected; see GM2416 Fibroblast
Diabetes Mellitus: Maturity Onset Diabetes - 22210											
GM1905		R1640 20I	0034	32	F			PROBAND			MODY diabetes diagnosed at age 10; both parents and three brothers had MOD diabetes; HLA type A11,-/Bw35,-
GM1247		R1640 20I	0034	29	M	WH		PROBAND	95	1	MODY; onset at age 12; see GM1237 Fibroblast
GM1246		R1640 20I	0034	46	M	WH		AFF BRO	95	2	MODY; onset at age 29; see GM1219 Fibroblast
GM1240		R1640 20I	0034	33	F	WH		AFF SIS	95	3	MODY; onset at age 16; see GM1122 Fibroblast

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus: Maturity Onset Diabetes - 22210											
GM1243		R1640	20I 0034	35	M	WH		AFF BRO	95	4	MODY; onset at age 16; see GM1430 Fibroblast
GM1956		R1640	20I 0034	44	M	WH		AFF BRO	95	6	MODY; onset at age 26; see GM1955 Fibroblast
GM1498		R1640	20I 0034		F	WH		AFF SIS	95	7	MODY; see GM1497 Fibroblast
GM1242		R1640	20I 0034	22	M	WH		AFF NEPH	95	8	MODY; onset at age 14; see GM1435 Fibroblast; son of GM1498
GM1244		R1640	20I 0034	20	M	WH		AFF NEPH	95	9	MODY; onset at age 12; see GM1496 Fibroblast; son of GM1498
*GM1410		R1640	20I 0034	15	F	WH		AFF NIECE	95	10	See GM1409 Fibro; mat cous of GM1242, 1244;onset age 13; only JOD in fam.; on Insul.;HLA type A2,Aw24,Bw35,B13(B12)
GM1838		R1640	20I 0034	22	F	WH		AFF COU	95	11	MODY type II; onset at age 18; pat.1st cousin once removed of the 6 sibs; HLA type A28(A2),Aw30,B12(B13),Bw35; see GM1837 Fibroblast
GM1241		R1640	20I 0034	37	M	WH		PROBAND	291	1	Third generation of MOD; HLA type A3,A10,B13,B12; see GM1486 Fibroblast
Diabetes Mellitus: Juvenile Onset - 22210											
GM2676		R1640	20I 0095	17	M	WH		PROBAND	236	1	Onset at age 2; on Insulin; HLA type A1,B8/A10,B18; see GM2637 Fibroblast
GM2793A		R1640	20I 0260	10	F	WH		AFF SIS	236	2	Onset at age 3; on Insulin; HLA type A1,B8/A10,B18; see GM2638 Fibroblast
GM2675		R1640	20I 0095	19	M	WH		BRO	236	3	See GM2639 fibroblast; HLA type A1, B8/A10,B18; non-diabetic
GM2741A		R1640	20I 0095	41	M	WH		FATHER	236	4	Non-diabetic; HLA type A1,B8/A3,Bw35; see GM2640 Fibroblast; both parents had maturity onset diabetes
GM3355		R1640	20U 0260	41	F	WH		MOTHER	236	5	Non-diabetic; HLA type A10(25),B18/A3,B7 see GM2641 Fibroblast
GM2713		R1640	20I 0095	17	M	WH		PROBAND	243	1	Onset at age 14; on Insulin; HLA type A1,B8/A9,B18; see GM2712 Fibroblast
GM2705		R1640	20I 0095	45	M	WH		FATHER	243	2	Non-diabetic; HLA type A1,B8/A2,Bw35; see GM2704 Fibroblast

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus: Juvenile Onset - 22210											
GM2707		R1640 20I	0095	45	F	WH		MOTHER	243	3	Non-diabetic; HLA type A9(24),B18/A2,B7; see GM2706 Fibroblast
GM2709		R1640 20I	0095	16	M	WH		BRO	243	4	Non-diabetic; HLA type A2,B7/A2,Bw35; see GM2708 Fibroblast
GM2711		R1640 20I	0095	10	M	WH		BRO	243	5	Non-diabetic; HLA type A2,B7/A2,Bw35; see GM2710 Fibroblast
GM2728		R1640 20I	0095	25	F	WH		SIS	243	6	Non-diabetic; HLA type A1,B8/A9,B18; see GM2869 Fibroblast
GM3027		R1640 20I	0260	23	M	WH		AFF BRO	243	8	Onset at age 11; HLA type A1,B8/A9,B18
GM3235		R1640 20I	0260	14	M	WH		PROBAND	606	1	HLA type A3,Bw17/A9(23),B12; 6 yrs old at onset; on Insulin; see GM3358 Fibroblast
GM3239		R1640 10U	0260	13	M	WH		BRO	606	2	HLA type A3,Bw17/A9(23),B12; non-diabetic; see GM3359 Fibroblast
GM3236		R1640 17U	0260	42	M	WH		AFF FATHER	606	3	HLA type A9(23),B12/A28,Bw15; onset at age 14 yrs; see GM3360 Fibroblast
GM3237		R1640 10U	0260	16	M	WH		BRO	606	4	HLA type A3,Bw17/A28,Bw15; non-diabetic; see GM3361 Fibroblast
GM3238		R1640 10U	0260	40	F	WH		MOTHER	606	5	HLA type A1,Bw40/A3,Bw17; non-diabetic; see GM3362 Fibroblast
GM2762		R1640 20I	0095	13	F	WH		PROBAND	620	1	Onset at age 10; on Insulin; HLA type A2,Bw16/A1,B8; see GM1872 Fibroblast
GM2763		R1640 20I	0095	17	F	WH		AFF SIS	620	2	Onset at age 13; on Insulin; HLA type A10,Bw16/A1,B8; see GM1873 Fibroblast
GM2765		R1640 20I	0095	21	M	WH		AFF BRO	620	3	Onset at age 4; on Insulin; HLA type A10,Bw16/A1,B8; see GM1909 Fibroblast; hypertension; short stature
GM3354A		R1640 20U	0260	21	F	WH		SIS	620	4	Non-diabetic; HLA type A2,Bw16/A3,B7; see GM1910 Fibroblast
GM2764		R1640 20I	0095	22	M	WH		AFF BRO	620	5	Onset at age 4; on Insulin; HLA type A2,Bw16/A1,B8; see GM1911 Fibroblast
GM2866A		R1640 20I	0095	19	M	WH		BRO	620	6	Non-diabetic; HLA type A2,Bw16/A1,B8; see GM1875 Fibroblast

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus: Juvenile Onset - 22210											
GM2766		R1640 20I	0095	24	F	WH		SIS	620	7	HLA type A2,Bw16/A1,B8; mentally retarded non-diabetic; see GM1874 Fibroblast
GM2812A		R1640 10U	0260	19	M	WH		BRO	620	8	Non-diabetic; HLA type A10,Bw16/A1,B8; see GM1876 Fibroblast
GM2714		R1640 20I	0095	42	F	WH		MOTHER	620	9	Non-diabetic; HLA type A1,B8/A3,B7; see GM1878 Fibroblast
GM3356A		R1640 10U	0260	53	M	WH		FATHER	620	10	Non-diabetic; HLA type A2,Bw16/A10,Bw16; see GM1877 Fibroblast
GM2677		R1640 20I	0095	24	F	WH		PROBAND	621	1	Onset at age 6; HLA type A1,B8/ Aw31, Bw40; see GM2613 Fibroblast
GM2678		R1640 20I	0095	27	M	WH		AFF BRO	621	2	Onset at age 18 mo.; on Insulin; HLA type A1,B8/A1,--; see GM2614 Fibroblast
GM2679		R1640 20I	0095	29	M	WH		BRO	621	3	See GM2615 Fibroblast; HLA type A1, B8/A1,--; non-diabetic
GM2680		R1640 20I	0095	63	M	WH		FATHER	621	4	Non-diabetic; HLA type A1,B8/A2,Bw15; see GM2616 Fibroblast
GM2785		R1640 20I	0095	60	F	WH		MOTHER	621	5	Non-diabetic; HLA type Aw31,Bw40/A1,--; see GM2617 Fibroblast
Diabetes Mellitus and Insipidus with Optic Atrophy - 22230											
GM1795		R1640 20I	0189	13	F	WH		PROBAND	96	1	See GM1610 Fibroblast; HLA type Aw32, B12/Aw32,Bw40; on Insulin; onset at age 11; optic atrophy
GM1799		R1640 20I	0189	18	F	WH		AFF SIS	96	2	See GM1609 Fibroblast; HLA type Aw32, B12/A9,Bw35; not on Insulin; onset at age 16; optic atrophy
GM1797		R1640 20I	0189	42	M	WH		FATHER	96	4	Non-diabetic; HLA type A9,Bw35/Aw32, Bw40; see GM1701 Fibroblast
GM1796		R1640 20I	0189	16	M	WH		BRO	96	5	Non-diabetic; HLA type Aw32,Bw40/ Aw32,B12
GM1800		R1640 20I	0189	7	F	WH		SIS	96	6	Non-diabetic twin of GM1801; HLA type A2,Bw15/A9,Bw35
GM1801		R1640 20I	0189	7	F	WH		SIS	96	7	Non-diabetic twin of 1800; HLA type A2, Bw15/A9,Bw35

HUMAN LYMPHOCYTE CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus and Insipidus with Optic Atrophy - 22230											
GM1930		R1640 20U	0189	39	F	WH		MOTHER	96	8	Non-diabetic; HLA type A2,Bw15/Aw32,B12; formerly GM1802
GM1803		R1640 20I	0189	11	F	WH		SIS	96	9	Non-diabetic; HLA type Aw32,B12/A9,Bw35
Dysautonomia (Riley-Day Syndrome) - 22390											
GM0599		R1640 20I	0024		F	WH	--	PROBAND			
GM0600		R1640 20I	0024		M	WH	+-(0)	PARENT			
GM0601		R1640 20I	0024	10	M	WH	--	PROBAND			
GM0602		R1640 20I	0024		M	WH	+-(0)	PARENT			
GM0603		R1640 20I	0024		F	WH	+-(0)	PARENT			
GM1465		R1640 20I	0008	21	F	WH	--	PROBAND			
GM1466		R1640 20I	0008	11	M	WH	--	PROBAND			
GM1777		R1640 20I	0008	22	F	WH	--	PROBAND			
Dyskeratosis Congenita - 30500											
GM1775		R1640 20I	0107	7	M	WH	y-	PROBAND	99	1	See GM1774 Fibroblast; HLA type Aw30,(Bw40),B13,B5; maternal grandfather was affected
*GM3193		R1640 20I	0295	15	M	WH	y-	PROBAND	515	1	
*GM3194		R1640 20I	0295	18	M	WH	y-	AFF BRO	515	2	
*GM3195		R1640 20I	0295	19	M	WH	y-	AFF BRO	515	3	
*GM3650		R1640 20I	0295	47	F	WH	+-(0)	MOTHER	515	4	Clinically normal
Huntington's Disease - 14310											
GM2080		R1640 20I	0136	48	F	WH	+-	PROBAND	107	1	46,XX; see GM2079 Fibroblast; father is also affected
GM2078		R1640 20I	0136	25	F	WH	+?	DAU	107	2	At risk (50%); see GM2077 Fibroblast
GM2146		R1640 20I	0186	54	M	WH	+-	PROBAND	108	1	Mother was also affected; see GM2147 Fibroblast
GM2148		R1640 20I	0186	54	F	WH	++	SPOUSE	108	2	Normal wife; see GM2149 Fibroblast
GM2150		R1640 20I	0186	26	F	WH	+?	DAU	108	3	At risk (50%); see GM2151 Fibroblast; personality disorder but no chorea

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Huntington's Disease - 14310											
GM2152		R1640 20I	0186	40	F	WH	++	SPOUSE	109	2	Normal widow of proband; see GM2153 Fibroblast
GM2154		R1640 20I	0186	20	F	WH	+?	DAU	109	3	At risk (50%); see GM2155 Fibroblast
GM2156		R1640 20I	0186	16	F	WH	+?	DAU	109	4	At risk (50%); see GM2157 Fibroblast
GM2158		R1640 20I	0186	19	F	WH	+?	DAU	109	5	At risk (50%); see GM2159 Fibroblast
GM2160		R1640 20I	0186	21	M	WH	+?	SON	109	6	At risk (50%); see GM2161 Fibroblast
GM2162		R1640 20I	0186	12	M	WH	+?	SON	109	7	At risk (50%); see GM2163 Fibroblast
GM2176		R1640 20I	0186	26	M	WH	+?	SON	110	2	At risk (50%); see GM2177 Fibroblast
GM2182		R1640 20I	0186	21	F	WH	+?	DAU	110	3	At risk (50%); see GM2183 Fibroblast
GM2168		R1640 20I	0186	52	M	WH	++	BRO-IN-LAW	110	4	Normal spouse of GM2166; see GM2169 Fibroblast
GM2186		R1640 20I	0186	60	F	WH	+?	SIS	110	5	Asymptomatic at age 60; reduced at risk; see GM2187 Fibroblast
GM2188		R1640 20I	0186	63	M	WH	++	BRO-IN-LAW	110	6	Normal spouse of GM2186; see GM2189 Fibroblast
GM2166		R1640 20I	0186	58	F	WH	+-	AFF SIS	110	7	46,XX/47,XX,+12; (74%/26%); proband for this family is GM2165 Fibroblast; see GM2167A Fibroblast
GM2178		R1640 20I	0186	38	F	WH	+?	NIECE	110	8	Daughter of reduced at risk GM2186; reduced at risk
GM2180		R1640 20I	0186	51	F	WH	++	SPOUSE	110	9	Normal
GM3620		R1640 20I	0186	29	F	WH	+-	AFF DAU	110	10	See GM3621 Fibroblast; age of onset 18 years
GM2172		R1640 20I	0186	52	F	WH	+-	PROBAND	111	1	See GM2173 Fibroblast
GM2170		R1640 20I	0186	22	F	WH	+?	DAU	111	2	At risk (50%); see GM2171 Fibroblast
GM2174		R1640 20I	0186	55	M	WH	++	SPOUSE	111	3	See GM2175 Fibroblast
GM2190		R1640 20I	0190	32	F	WH	+?	MOTHER	206	2	At risk (50%); mother of GM2123 Fibroblast; see also GM2191 Fibroblast
GM3642		R1640 15I	0136	55	M	WH	+-	PROBAND	292	1	See GM1061 Fibroblast; choreoathetosis, dysarthria, dysphagia; positive family history for H.D.

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Huntington's Disease - 14310												
GM3644		R1640 15I	0136	48	M	WH	+-		PROBAND	293	1	See GM1085 Fibroblast; moderate ataxia, chorea, dysarthria, & dysphagia; onset at age 35; affected mother & grandmother
GM3643		R1640 15I	0136	50	F	WH	+-		AFF SIS	293	2	See GM1083A Fibroblast; onset at age 35; choreoathetosis, mild dysarthria, and mild dysphagia
GM3645		R1640 20I	0136	53	F	WH	+-		AFF SIS	293	3	Severe choreoathetosis; moderate dysarthria and dysphagia; onset at age 33
GM3745		R1640 20I	0136	22	F	WH	+?		DAU	293	4	At risk (50%)
GM3746		R1640 20I	0136	24	F	WH	+?		DAU	293	5	At risk (50%)
GM3747		R1640 20I	0136	18	M	WH	+?		SON	293	6	At risk (50%)
GM3748		R1640 20I	0136	33	M	WH	+?		NEPH	293	7	Son of GM3645; at risk (50%)
Hyper IgM												
GM0390		R1640 20I	0010	3	F	WH			PROBAND			46,XX
Leukemia, Sub-acute												
GM2584		R1640 20I	0008	60	M	WH		A	PROBAND			Culture has 3 populations of cells: lymphoblasts, leukemic, & plasma cells; leukemic are esterase positive (T-cell leukemia)
Mental Retardation, X-Linked - 30950												
*GM3200		R1640 20I	0061	34	M	WH	y-		PROBAND			46,fra(X)(q27),Y; 17% in peripheral blood leukocytes; 46,XY in lymphoid culture
Multiple Myeloma - 25450												
*GM1500		R1640 20I	0010		M				PROBAND			IgG Myeloma; IgG+,Lambda+; HLA type A2(A3),B12,Bw15
*GM2132		R1640 20I	0205	61	M				PROBAND			IgG Lambda-type light chain secreting: RPMI 8226; ATCC CCL 155
GM1311		R1640 20I	0051	68	M	BL			PROBAND	180	1	Bone marrow; HLA type A10,A11(A3),B12
GM1312		R1640 20I	0051	68	M	BL			SAME	180	1	Peripheral blood; HLA type A10,A11(A3), B12
Neurofibromatosis (von Recklinghausen Disease) - 16220												
GM1634		R1640 20I	0104	61	M	WH	+-		PROBAND	113	1	Atypical; see GM1633 Fibroblast

HUMAN LYMPHOCYTE CULTURES

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Neurofibromatosis (von Recklinghausen Disease) - 16220											
GM1641		R1640 20I	0104	19	F	BL	+-	PROBAND	114	1	See GM1639 Fibroblast
GM1861		R1640 20I	0104	41	M	WH	+-	PROBAND	115	1	See GM1860 Fibroblast; HLA type A3,A2, B27,Bw22(Bw40)
GM3421		R1640 20I	0294	39	F	WH	+-	AFF MOTHER	212	1	
GM3420		R1640 20I	0294	17	F	WH	+-	PROBAND	212	2	Cafe-au-lait spots; neurofibromas; mental retardation
Olivopontocerebellar Atrophy - 16440, 50, 60, 70, and 25830											
GM3562		R1640 20I	0110	22	F	BL		PROBAND	240	1	Probably type III; see GM3561 Fibroblast; positive family history
Polyposis, Intestinal: Type III (Gardner Syndrome) - 17530											
GM2633		R1640 20I	0225	31	F	WH	+-	PROBAND	522	1	Many affected members in pedigree
GM3224		R1640 20I	0308	25	M	WH	+-	AFF SON	603	1	See GM3223 Fibroblast
GM3315A		R1640 20I	0308	48	F	WH	+-	PROBAND	603	2	See GM3314 Fibroblast; adenocarcinoma of colon
Rothmund-Thomson Syndrome - 26840											
GM2451		R1640 20I	0155	25	M	WH	--	PROBAND			Clinically affected
Schizophrenia and Psychiatric Disorders - 18150											
GM1793		R1640 20I	0137	26	M	WH		PROBAND	119	2	See GM1792 Fibroblast; HLA type Aw24, B18,B13; episodes of agitation, delusions of persecution, fear of assassination; father also affected
GM1836		R1640 20I	0137	27	F	WH		AFF SIS	119	3	See GM1835 Fibroblast; HLA type A28, Bw16/A3,Bw35; hospitalized; drugs; father also affected
GM1488		R1640 20I	0137	23	M	WH		PROBAND	176	1	HLA type Aw26,B16/Aw26,B16; see GM2497 Fibroblast; paralogical thinking, affective shielding, splitting of affect from content, suspiciousness
GM1487		R1640 20I	0137	53	F	WH		MOTHER	176	2	Unaffected; HLA type Aw26,B16/A2,B35 described as "unreasonable and demanding" by family members
GM1489		R1640 20I	0137	55	M	WH		AFF FATHER	176	3	HLA type Aw26,B16/A18,B-; 3 episodes of depression; ECT; no hypomania
GM1490		R1640 20I	0137	27	F	WH		AFF SIS	176	4	HLA type Aw26,B16/A2,B35; see GM2503 Fibroblast; anorexia nervosa since adolescence; more schizoid than depressed

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Tuberous Sclerosis - 19110											
GM3549		R1640 20I	0006	16	M	WH	+-	PROBAND			Positive family history
GM1636		R1640 20I	0104	17	M	BL	+-	PROBAND	124	1	See GM1643 and GM1635 Fibroblast
GM1638		R1640 20U	0104	20	F	BL	+-	PROBAND	125	1	See GM1644 Fibroblast
Waldenstrom Macroglobulinemia - 15360											
GM1501		R1640 20I	0010	69	M		+-	PROBAND			HLA type A3,A9,Bw22,(B12)
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE											
Ataxia-Telangiectasia - 20890											
GM0717		R1640 20I	0066	11	M		--	PROBAND			
GM0719		R1640 20I	0066	13	F	WH	--	PROBAND	177	1	46,XX
*GM0718		R1640 20I	0066	9	M	WH	--	AFF BRO	177	2	
GM0736		R1640 20I	0066		M	WH	+-(O)	FATHER	177	3	46,XY
GM0781		R1640 20I	0066	4	F	WH	--	AFF SIS	177	4	
*GM1526		R1640 20I	0054	28	F	WH	--	PROBAND	178	1	46,XX,t(X;14) clone in short term lymphocytes; HLA type A9,A10,Bw17, B13(Bw40)
*GM1525		R1640 20I	0054	19	F	WH	--	AFF SIS	178	2	46,XX,t(14;14) clone in short term lymphocytes; HLA type A28,(Aw31,33),B12
GM2782		R1640 20I	0077	5	M	WH	--	PROBAND	228	1	Iranian; same patient as GM2532; see GM2531 Fibroblast
GM2783		R1640 20I	0077	8	M	WH	--	AFF BRO	228	2	Iranian; see GM2530 Fibroblast
GM2781		R1640 20I	0077	40	F	WH	+-(O)	MOTHER	228	3	Iranian; clinically normal
GM3187		R1640 20I	0226	40	M	WH	+-(O)	FATHER	516	1	Clinically normal; see GM3488 Fibroblast
GM3188		R1640 20I	0226	36	F	WH	+-(O)	MOTHER	516	2	Clinically normal; see GM3489 Fibroblast
GM3189		R1640 20I	0226	7	M	WH	--	PROBAND	516	3	Neurological symptoms; see GM3487 Fibroblast
GM3324		R1640 20I	0226	15	F	WH	+?	SIS	516	4	Possible heterozygote; clinically normal; see GM3490 Fibroblast

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SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE												
Ataxia-Telangiectasia - 20890												
GM3323		R1640 20I	0226	6	M	WH	+?		BRO	516	5	Possible heterozygote; clinically normal; see GM3492 Fibroblast
GM3325		R1640 20I	0226	14	F	WH	+?		SIS	516	6	Possible heterozygote; clinically normal; see GM3491 Fibroblast
GM3332		R1640 20I	0226	13	M	BL	--		PROBAND	605	1	See GM3395 Fibroblast; a deceased sister had A.T.
GM3382		R1640 20I	0226	43	M	BL	+-(0)		FATHER	605	2	See GM3397 Fibroblast; clinically normal
GM3334		R1640 20I	0226	42	F	BL	+-(0)		MOTHER	605	3	See GM3396 Fibroblast; clinically normal
GM3333		R1640 20I	0226	11	F	BL	+?		SIS	605	4	Normal; possible heterozygote; see GM3400 Fibroblast
GM3380		R1640 20I	0226	20	F	BL	+?		SIS	605	5	Normal; possible heterozygote; see GM3399 Fibroblast
GM3383		R1640 20I	0226	22	M	BL	+?		BRO	605	6	Normal; possible heterozygote; see GM3398 Fibroblast
LYMPHOCYTE CHROMOSOME ABERRATIONS: DELETIONS												
Deletion												
GM2054		R1640 20I	0194	3	M	WH		A	PROBAND			46,XY,del(18)(pter>q21:)
GM3470		R1640 20I	0120	1	F	WH			PROBAND	239	2	46,XX,del(10)(p12p13); microcephaly, microophthalmia, and hypotelorism; father is GM3469 Lymphoid; both parents are chromosomally normal
Iso X Chromosome												
GM3543		R1640 20I	0107	15	F	WH			PROBAND			Turner Syndrome; 46,X,i(Xq)
LYMPHOCYTE CHROMOSOME ABERRATIONS: TRANSLOCATIONS												
Translocation												
*GM0633		R1640 20I	0089	8	M	WH		A	PROBAND			46,XY,t(7;10)(7qter>7p21::10q11>10qter; 10pter>10q11::7p21>7pter)
GM1201		R1640 20I	0144	14MO	F	WH		B	PROBAND			46,XX,-21,+t(21;21) in peripheral blood of a Down's syndrome patient
GM1203		R1640 20I	0066	30	F	WH		B	PROBAND			46,XX,t(4;12)

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LYMPHOCYTE CHROMOSOME ABERRATIONS: TRANSLOCATIONS												
Translocation												
*GM1261		R1640 20I	0107	13	F	WH		A	PROBAND			45,XX,-13,-18,+t(13;18)(13qter>cen>18qter); HLA type A1,A2,Bw15(B18); 18p- syndrome
GM2669		R1640 20I	0039	27	M	WH		A	PROBAND			46,XY,t(6;8)(6pter>6q27::8q21>8qter;8pter>8q21::6q27>6qter)
GM3423		R1640 20I	0058	32	F	WH		B	PROBAND			46,XX,t(1;6)(p36;p21); balanced; Puerto Rican
GM3559		R1640 20I	0328	30	M	WH			PROBAND			46,XY,t(15;2)
GM2324		R1640 20I	0182	27	F	WH		A	MOTHER	179	2	46,XX,t(16;22)(16qter>16p13::22q12>22qter;22pter>22q21::16p13>16pter),9qh+; mother of GM2325 Fibroblast
GM2820		R1640 20I	0107	7MO	F	BL		B	PROBAND	324	1	46,XX,-12,der(12)t(9;12)(p13;q24)mat;see GM2819 Fibroblast
GM2822		R1640 20I	0107		F	BL		B	MOTHER	324	2	46,XX,t(9;12)(p13;q24); see GM2821 Fibroblast
*GM1388		R1640 20I	0061	14	F	WH		B	PROBAND	369	1	46,XX,-13,+der(9),t(9;13)(q22;q12)mat; see GM1387 Fibroblast
GM1561		R1640 20I	0008	36	F	WH		B	PROBAND	397	1	46,XX,t(4;11)(q25;q13); HLA type A10,Aw24,Bw35,(B12)
GM3197		R1640 20I	0318	31	F	WH		B	PROBAND	518	1	46,XX,t(17;22)(p13;q11); see GM3196 Fibroblast
GM3316		R1640 20I	0311	3	F	WH		A	PROBAND	617	1	46,X,t(X;11)(Xpter>Xq11:11p11>11pter;11qter>11p11::Xp11>Xqter); see GM2859 Fibroblast; South American
*GM1063A		R1640 20I	0067	12	F	BL		A	PROBAND	638	1	46,XX,t(4;13)(4pter>4q31::13q14>13qter;13pter>13q14::4q31>4qter);karyotype unstable; HLA type A1,B12,Bw17; see GM0972 Fibroblast
LYMPHOCYTE CHROMOSOME ABERRATIONS: ANEUPLOIDY												
Polysomy												
*GM1202		R1640 20I	0066	7	M	WH		A	PROBAND			49,XXXXY
GM3384		R1640 20I	0077	9	M	WH		B	PROBAND			Arab; 49,XXXXY
GM1416		R1640 20I	0061	27	F	WH		A	PROBAND	370	1	48,XXXX; see GM1415 Fibroblast

HUMAN LYMPHOCYTE CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
LYMPHOCYTE CHROMOSOME ABERRATIONS: ANEUPLOIDY												
Trisomy												
GM3716		R1640 15I	0343	23	M	WH			PROBAND			Down's syndrome
*GM1921		R1640 20I	0061	23	M	WH		B	PROBAND	418	1	47,XY,+21,inv(9)(p13q13)mat; see GM1920 Fibroblast; Down's syndrome patient
SPECIALLY CHARACTERIZED HUMAN LYMPHOCYTE CULTURES												
HLA Histocompatability Type - 14280 - 14286												
GM3096		R1640 20I	0300					B	PROBAND			HLA type: A:2,B:15,C:3,D:4 Bodmer I.D.; JHH
GM3097		R1640 20I	0300					B	PROBAND			HLA type: A:26,26,B:16,16,D:10,10 Bodmer I.D.; FSH
GM3098		R1640 20I	0300					B	PROBAND			HLA type; A:2,2,B:17,17,DR:3,3 Bodmer I.D.; WT49
GM3099		R1640 20I	0300					B	PROBAND			HLA type:A:2,-,B:35,-,C:4,-,D:1,1,DR:1,1 Bodmer I.D.; MAJA
GM3103		R1640 20I	0300					B	PROBAND			HLA type; A:9,9,B:14,14,DR:4,4 Bodmer I.D.; WT51
GM3104A		R1640 20I	0300					B	PROBAND			HLA type; A:3,3,B:35,35,C:4,4,D:1,1, DR:1,1; Bodmer I.D.; IBW4
GM3105A		R1640 20I	0300					B	PROBAND			HLA type; A:26,28,B:38,18,D:5,DR;5 Bodmer I.D.; IDF
GM3106		R1640 20I	0300					B	PROBAND			HLA type; A:1,B:35,C:4 Bodmer I.D., FPF
GM3107		R1640 20I	0300					B	PROBAND			HLA type; A:3,B:7,D:2,DR:2 Bodmer I.D.; PGF
GM3158		R1640 20I	0300					B	PROBAND			HLA type; A:31,B;16,D:5 Bodmer I.D.; DHF
GM3160		R1640 20I	0300					B	PROBAND			HLA type; A:2,2,B:35,12,DR:4x7 Bodmer I.D.; HERLUFF
GM3161		R1640 20I	0300					B	PROBAND			HLA type; A:3,3,B:7,7,D:2,2,DR:2,2 Bodmer I.D.; MST1
GM3162		R1640 20I	0300					B	PROBAND			HLA type; A:2,B:8,35,D:3,DR:3 Bodmer I.D.; MWF
GM3163		R1640 20I	0300					B	PROBAND			HLA type; A:30,B:13,C:2,DR:7 Bodmer I.D.; LBF

HUMAN LYMPHOCYTE CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
SPECIALLY CHARACTERIZED HUMAN LYMPHOCYTE CULTURES												
HLA Histocompatability Type - 14280 - 14286												
GM3164		R1640 20I	0300					B	PROBAND			HLA type; A:2,24,B:27,C:2,D:4,DR:4,4 Bodmer I.D.; AS
GM3190		R1640 20I	0300					B	PROBAND			B cell line, no HLA:A,B, or C; no B2 Microglobulin, HLA; D:6; Bodmer I.D.; DAUDI
GM3357		R1640 10I	0300					B	PROBAND			HLA type A:1,2,B:8,13,C:2?,DR:2 Bodmer I.D.; Bristol-8

HUMAN AMNIOTIC FLUID CELL CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
BIOCHEMICAL MUTANT CONDITIONS: HUMAN AMNIOTIC FLUID CELLS											
Cystinosis: Early Onset (Nephropathic Type; Infantile Type) - 21980											
GM0804		EEMEM	20U 0121	22FW	M	WH	-- B	PROBAND			46,XY; increased intracellular level of 1/2 cystine; passage 3 at IMR
Fabry Disease (Diffuse Angiokeratoma) - 30150											
GM0636	8	HEMEM	20U 0017	18FW	M	WH	y- B	PROBAND			46,XY; Alpha-galactosidase deficient
Galactosemia (Transferase Deficiency) - 23040											
GM1743	3	HF12	20U 0077	24FW	M	WH	-- B	PROBAND	32	1	46,XY; see GM1741 Fibroblast
Homocystinuria (Cystathionine Synthetase Deficiency) - 23620											
GM0919	6	EEMEM	20U 0038	30FW	F		-- B	PROBAND			46,XX; deficient enzyme activity; mother is also affected
Hypophosphatasia - 24150											
GM3336		EEMEM	20U 0321	20FW	F	WH	-- B	PROBAND	604	1	46,XX; severe in utero onset form; deficient alkaline phosphatase; passage 5 at IMR; parents are GM3327 & GM3328 Fibroblasts; see also GM3502 Fibroblast
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800											
GM0236	13	HF10	20U 0085	16FW	M		y- B	PROBAND			HGPRT deficient
*GM2338	5	EEMEM	20U 0183	15FW	M	WH	y- A	PROBAND	74	1	46,XY; see GM2290 and GM2291 Fibroblast and GM2292 Lymphoid; two similarly affected male sibs and a maternal uncle
Metachromatic Leukodystrophy: Infantile - 25010											
GM2095		HF12	20U 0105	18FW	F	AI	--	PROBAND			Passage 3 at IMR; after delivery showed increased urinary sulfatides
GM2214		HF12	20U 0105	20FW	M	WH	-?	PROBAND			Passage 4 at IMR; sister has typical infantile MLD
Mucolipidosis: Type IV - 25265											
*GM2529	19	HF12	20U 0077	19FW		WH	-- B	AFF SIB	229	2	Ashkenazi; abnormal cytoplasmic lysosomal storage bodies; affected sib is GM2048 Fibroblast & GM2533 Lymphoid
Mucopolysaccharidosis: Type II - Hunter Syndrome - 30990											
GM1584	6	HF12	20U 0077	21FW	M	WH	y- A	AFF BRO	247	2	46,XY; deficient in iduronidase sulfatase; Israeli; affected brother is GM1583 Fibroblast
CHROMOSOMAL ABERRATIONS: HUMAN AMNIOTIC FLUID CELLS											
Aneuploid											
GM1993	7	MC5A	20U 0172	21FW	M	WH	A	PROBAND			47,XYY

HUMAN AMNIOTIC FLUID CELL CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
CHROMOSOMAL ABERRATIONS: HUMAN AMNIOTIC FLUID CELLS											
Aneuploid											
GM2269	9	EEMEM	20U 0109	18FW	M	WH	A	PROBAND			47,XXY
GM2419	7	MC5A	20U 0172	16FW	F	WH	B	PROBAND			47,XX,+21
GM2422	4	MC5A	20U 0172	16FW	F	WH	B	PROBAND			46,XX/47,XX,+18; 50%/50%
GM2587	5	MC5A	20U 0172	17FW	M	WH	A	PROBAND			47,XYY
GM3090		MC5A	20U 0172	23FW	F	WH	A	PROBAND			Bilateral simian crease; incurving fifth finger; dislocated hip; 46,XX,21q+; passage 2 at IMR
GM3250	7	MC5A	20U 0172	15FW	M	WH	A	PROBAND			47,XY,+18
GM3416	7	MC5A	20U 0172	16FW	F	OR	A	PROBAND			47,XXX
GM3535	5	HF12	20U 0172	20FW	M	WH	B	PROBAND			47,XXY
GM3606	4	HF12	20U 0172	18FW	F	OT	B	PROBAND			Mulatto; 47,XX,+21
GM3673	7	HF12	20U 0342	19FW	F	BL	A	PROBAND			47,XX,+del(9)(pter>q21:)mat
GM3686	5	MC5A	20U 0172	18FW	F	WH	B	PROBAND			Hispanic; 47,XX,+21
GM3699	5	EEMEM	20U 0172	18FW	F	IN	B	PROBAND			47,XXX
GM3769	5	EEMEM	20U 0172	16FW	M	WH	B	PROBAND			47,XY,+18
GM3091	4	MC5A	20U 0172	15FW	M	WH	A	PROBAND	448	1	47,XXY; see GM3102 Fibroblast
Deletion											
GM2421	6	MC5A	20U 0172	16FW	F	WH	B	PROBAND			46,XX,13p-(mat)
GM3202	8	MC5A	20U 0172	17FW	F	WH	B	PROBAND			46,XX/46,XX,del(X)(qter>p11:)
Inversion											
GM2055	7	MC5A	20U 0172	17FW	F		B	PROBAND			46,XX; inv(9); father shows same inv(9) in blood; family history of spontaneous abortions and infertility
GM3367	7	MC5A	30I 0172	16FW	F	OR	B	PROBAND			46,XX,inv(18)pat; balanced
Marker Chromosome											
GM2662	6	MC5A	20U 0172	21FW	F	BL	A	PROBAND			47,XX,+mar; marker is denovo and satellited
Translocation											
GM0477	9	EEMEM	20U 0048	14FW	F	WH	B	PROBAND			46,XX,t(13;18) with trisomy 13q

HUMAN AMNIOTIC FLUID CELL CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
CHROMOSOMAL ABERRATIONS: HUMAN AMNIOTIC FLUID CELLS												
Translocation												
GM3435		HF12	20U 0273	15FW	M			B	PROBAND			46,XY,t(4;11)(p14;p15)der(4)mat; passage 3 at IMR
GM3100	8	MC5A	20U 0172	18FW	F	WH		A	PROBAND	449	1	46,XX,t(9;17)(9qter>9p22::17p11>17pter; 17qter>17p11::9p22>9pter)mat; mother is GM3119 Fibroblast
APPARENTLY NORMAL: HUMAN AMNIOTIC FLUID CELLS												
Apparently Normal (Amniotic)												
GM0472	9	R1640	30I 0058	19FW	M			B	PROBAND			46,XY
GM0473	6	R1640	30I 0058	17FW	M			A	PROBAND			46,XY
GM0474	10	R1640	30I 0058	18FW	M			A	PROBAND			46,XY
GM0956	4	R1640	20U 0064	17FW	F			B	PROBAND			46,XX
GM0957	3	R1640	20U 0064	18FW	F			B	PROBAND			46,XX
GM1420	11	EEMEM	20U 0109	18FW	F	BL		B	PROBAND			46,XX (with long 1qh+)

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	VERIFIED	FAM#	FAMM#	REMARKS
Bovine Tissue							
GM3655	2	HF12	20U	0336			Skin biopsy of a 5 year old apparently normal Holstein cow
Canine Tissue							
GM0757	8	MC5A	20U	0095			Skin fibroblast culture; apparently normal female dog
GM0758	5	MC5A	20U	0095			6 mo. old female dog with Krabbe's Globoid Leukodystrophy; fibroblast culture
Chinese Hamster							
*GM0215		EEMEM	05U	0022			Line V79; young adult male, estab. by Ford and Yerganion, fibroblast culture established from lung biopsy
*GM0458		EEMEM	10U	0053			Line CHW, male, fibroblast cell line estab. by spontaneous transformation of a normal skin culture
*GM0459	2	EEMEM	10U	0053	A		Line CHW-1102, MMS induced mutant of GM0458, selected with 8-azaguanine, HGPRT deficient; fibroblast culture
Syrian Baby Hamster Kidney							
*GM0345		EEMEM	10U	0083	B		Line TG-2, HGPRT deficient, selectable with 6-thioguanine, heteroploid, fibroblast culture
*GM0348		EEMEM	10U	0083	B		Line B-1, fibroblast culture, thymidine kinase deficient, selectable with 5-bromodeoxyuridine, heteroploid
*GM0511		EEMEM	10U	0083	B		Line A-5, subclone of B-1(GM0348), increased folate reductase, thymidine kinase deficient; fibroblast culture; heteroploid
Mouse Tissue							
*GM0086		EEMEM	15U	0040	A		Friend erythroleukemic cell line, clone #745, DMSO inducible, derived from DBA/2 mouse, grows in suspension
*GM0346		EEMEM	10U	0083	A		Line A-9, mouse L cell, fibroblast-like, HGPRT deficient, selectable with 8-azaguanine, heteroploid
*GM0347		EEMEM	10U	0083	A		Line B-82, mouse L cell, fibroblast-like, thymidine kinase deficient, sel. with 5-bromodeoxyuridine, heteroploid

ANIMAL CELL CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	VERIFIED	FAM#	FAMM#	REMARKS
Mouse Tissue							
*GM0979		HF12	20I 0158	A			Murine erythroleukemic line, T3-C12, derived from a splenic focus in DDD Mouse infected with Friend Leukemia virus; suspension culture
Ovine Tissue							
GM3550	2	HF12	20U 0325				Southdown sheep; female; 4 months old; fibroblast culture
Nonhuman Primates							
GM3442	3	HF12	20U 0325		535	1	Stumptail; new world female monkey; fibroblast culture from skin biopsy
GM3444	2	HF12	20U 0325		536	1	Stumptail; new world male monkey; fibroblast culture from skin biopsy
GM3446	2	HF12	20U 0325		537	1	Stumptail; new world male monkey; fibroblast culture from skin biopsy
GM3448	3	HF12	20U 0325		538	1	Chimpanzee; old world monkey; 7 yr old male; fibroblast culture from skin biopsy
GM3450	4	HF12	20U 0325		539	1	Chimpanzee; old world monkey; 15 yr old male; fibroblast culture from skin biopsy
GM3452	3	HF12	20U 0325		540	1	Chimpanzee; old world male monkey; fibroblast culture from skin biopsy
Myeloma Cell Lines for Hybridomas							
*GM3569		DUMEM	10U 0189	B			Mouse myeloma, line SP2/0; HPRT deficient; non-immunoglobulin producing
*GM3570		DUMEM	10U 0189	B			Mouse myeloma, line P3X63Ag8653, HPRT deficient; non-immunoglobulin producing; derived from P3 by Rajewsky
*GM3571		DUMEM	10U 0189	B			Mouse myeloma, line P3X63Ag8, HPRT deficient; secretes IgG1(K)(MOPC21), original Milstein line
GM3572		DUMEM	10U 0189	B			Mouse myeloma; line 45.6TG1.7; HPRT deficient; produces IgG2(K), derived by Scharff
GM3573		DUMEM	10U 0189	B			Mouse myeloma, line P3X63NS1, HPRT deficient, produces Kappa, non-immunoglobulin secreting

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	VERIFIED	FAM#	FAMM#	REMARKS
Hybridomas							
GM3565		DUMEM 20U	0339	A			Rat-mouse hybridoma; line J11d; produces IgM, Kappa; antigen specificity: Mouse thymocytes and peripheral B cells
*GM3566		DUMEM 20U	0339	A			Rat-mouse hybridoma; line D4.37; produces rat IgM, "non-Kappa"; mouse IgGp, Kappa; antigen specificity:RT1-N.A
*GM3568		DUMEM 20U	0339	A			Rat-mouse hybridoma; line D4.69; produces rat IgG2c, Kappa & mouse IgG1, Kappa; antigen specificity:RT1-N.A
*GM3704		DUMEM 20U	0339	A			Rat-mouse hybridoma; line PhK2.13.2; antigen specificity:RT1-N.A
*GM3705		DUMEM 20U	0339	A			Rat-mouse hybridoma; line PhN2.11.6; antigen specificity:RT1-N.A
*GM3710		DUMEM 20U	0339	A			Rat-mouse hybridoma; line PhR9.75.2; antigen specificity:RT1-N.A
*GM3713A		DUMEM 20U	0339	A			Rat-mouse hybridoma; line PhN2.34.1; antigen specificity:RT1-N.A

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AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
ANIMAL FIBROBLAST CULTURES												
Bovine Tissue												
AG2409		MC5A	20U	0241		M			PROBAND			Smooth muscle biopsy; abnormal polyploid cell line; 50-70% of the chromosomes appear to have Robertsonian translocations; passage one at IMR
AG2410		EEMEM	10U	0241		M			PROBAND			Smooth muscle biopsy; 60,XY; passage one at IMR
AG2411		MC5A	10U	0241		M			PROBAND			Smooth muscle biopsy; 60,XY; passage one at IMR
AG2791	1	EEMEM	20U	0281	4WK	M			PROBAND			Culture from pulmonary artery endothelium
Mouse Tissue												
AG2120	2	HF12	20U	0243					PROBAND			47,XO (Peromyscus ears); skin biopsy
HUMAN CELL CULTURES: AGING												
Alzheimer Disease of Brain - 10430												
AG0364	2	MC5A	20U	0139	53	M	WH	+-	PROBAND	87	1	Fibroblast culture
Ataxia-Telangiectasia - 20890												
AG2496	10	EEMEM	20U	0242	13	M		--	PROBAND			Skin fibroblast culture
AG2494	9	EEMEM	20U	0242	10	M		--	PROBAND	309	1	Skin fibroblast culture
AG2493	8	EEMEM	20U	0242	35	F		+-(O)	MOTHER	309	3	Skin fibroblast culture
*AG3058	4	HF12	20U	0296	14	F	BL	--	PROBAND	493	1	Also has acute lymphocytic leukemia; skin fibroblast culture
*AG3057	4	HF12	20U	0296	47	F	BL	+-(O)	MOTHER	493	2	Skin fibroblast culture
*AG3059	3	HF12	20U	0296	48	M	BL	+-(O)	FATHER	493	3	Skin fibroblast culture
Bloom Syndrome - 21090												
AG1620	22	EEMEM	20U	0044	18MO	F	WH	--	PROBAND	430	1	B.S. Registry #53(StAs); Line HG916; fibroblast culture
Central Core Disease of Muscle - 11700												
AG3728	3	EEMEM	20U	0348	53	M	WH	+-	PROBAND	529	1	Weakness & positive muscle biopsy - no type II fibers, type I fibers have central cores; fibroblast culture
Charcot-Marie Tooth Disease - 11820												
AG3758	3	HF12	20U	0348	49	F	WH	+-	PROBAND			Spinocerebellar degeneration; distal atrophy; hypercholesterolemia; skin fibroblast culture

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES: AGING

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
Cowden Disease - 15835												
AG1964	3	HF12	20U	0195	64	M	WH	++	PAT 1ST COU	181	1	46,XY,(9qh+); unaffected; fibroblast culture
AG1966	3	HF12	20U	0195	58	F	WH	+-	PROBAND	181	2	Benign tumors; tentative diagnosis: Cowdens; fibroblast culture; 46,XX; 40% of cells karyotyped showed chromatid breaks
AG1965	3	HF12	20U	0195	60	F	WH	++	MAT 1ST COU	181	3	Normal; fibroblast culture; 46,XX
AG1967	3	HF12	20U	0195		M	WH	++	PAT 1ST COU	181	4	Unaffected; fibroblast culture; 46,XY
AG1968	3	HF12	20U	0195	49	F	WH	++	PAT 1ST COU	181	5	Unaffected; fibroblast culture; 46,XX
AG1969	2	HF12	20U	0195		F	WH	+-	AFF NIECE	181	6	Cancer; tentative diagnosis:Cowdens; fibroblast culture
Cystic Fibrosis (Mucoviscidosis) - 21970												
AG3532	2	HF12	20U	0107	14	F	WH	--	PROBAND			Fibroblast culture from chest skin biopsy; hepatic failure
AG3533	2	HF12	20U	0107	23	M	WH	--	PROBAND			Fibroblast culture from chest skin biopsy; also juvenile diabetic on Insulin; pulmonary failure
AG3554	4	HF12	20U	0107	7MO	M	WH	--	PROBAND			Fibroblast culture from skin biopsy; pneumococcal meningitis; also has a sib affected with cystic fibrosis
AG3574	2	HF12	20U	0107	3MO	M	WH	--	PROBAND			Fibroblast culture from chest skin biopsy; diffuse alveolar disease
AG3556	3	HF12	20U	0107	NB	F	WH	--	PROBAND	544	1	Fibroblast culture from skin biopsy; stillborn; family history includes one affected cousin
Deletion												
*AG2719	6	EEMEM	20U	0244	29	M		A	PROBAND			Bilateral colobomas, multiple congenital anomalies of hands & feet; 46,XY,del(13)(pter>q22::q32>qter); fibroblast culture
Orthostatic Hypotension												
AG3633		R1640	20I	0363	66	M	WH		PROBAND	531	1	Dizziness; scotomata; syncope; no CNS manifestations; only affected family member; lymphoid culture
AG3634	4	HF12	20U	0363	59	M	WH		PROBAND	532	1	Dizziness; occasional syncope; impotence; anhidrosis; urinary frequency; only affected family member; fibroblast culture

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
HUMAN CELL CULTURES: AGING												
Orthostatic Hypotension												
AG3635		R1640 20I	0363	59	M	WH			PROBAND	532	1	Dizziness; occasional syncope; impotence; anhidrosis; urinary frequency; only affected family member; lymphoid culture
Lowe Oculocerebrorenal Syndrome - 30900												
AG1756	3	HF12 20U	0094	30MO	M		y-		PROBAND			Fibroblast culture
Mulibrey Nanism - 25325												
AG2122	7	MC5A 20U	0004	11	F		--		PROBAND			46,XX; fibroblast culture
AG2505	3	HF12 20U	0004	5	F	WH	--		PROBAND			Finnish; pre and postnatal growth failure; fibroblast culture
AG2506	2	HF12 20U	0004	9	F	WH	--		PROBAND			Finnish; growth failure, premature aging; fibroblast culture
Muscular Dystrophy; Duchenne Type (Pseudohypertrophic Progressive) - 31020												
AG3929		R1640 20I	0348	12	M	WH	y-		PROBAND			Lymphoblastoid culture; strong family history; affected brother and male cousins
Neuronal Atrophy												
AG3692	3	HF12 20U	0348	22	M	WH			PROBAND			Progressive motor neuropathy; distal and proximal weakness; evidence of denervation; fibroblast culture
Premature Aging: Unspecified												
AG3436		R1640 20I	0226	37	M	WH			PROBAND			46,XY/45,XO; premature aging; intestinal lymphangiectasia; cataracts; endocrine abnormalities; lymphoid culture
Progeria (See GM Repository) - 26410												
AG0998	15	EEMEM 20U	0055	20	M		--		PROBAND			Atypical; Cachectic Dwarfism; fibroblast culture
*AG0990	9	EEMEM 20U	0055		M		--		PROBAND			Atypical; also hypertriglyceridemia and abnormal glucose tolerance test; fibroblast culture; strain KH2(ATCC CRL1277)
*AG0991A	9	EEMEM 20U	0055	4	M		--		PROBAND			Classical; fibroblast culture; strain L.D.(ATCC CRL1277)
AG1177	8	EEMEM 20U	0049	9	M		--		PROBAND			46,XY; from Canadian Repository, WG360; fibroblast culture
AG1178	18	EEMEM 20U	0049	34	M		--		PROBAND			Slow growing fibroblast culture; from Canadian Repository, WG380

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
HUMAN CELL CULTURES: AGING											
Progeria (See GM Repository) - 26410											
AG1710	5	HF12	20U 0197		M		--	PROBAND			Atypical; progeria-like syndrome; fibroblast culture
*AG1972	8	HF12	20U 0091	14	F	WH	--	PROBAND			46,XX; classical; fibroblast culture
AG3506		R1640	20I 0309	13	M	WH	--	PROBAND	172	1	Mexican; lymphoblast culture
AG3513	3	HF12	20U 0309	13	M	WH	--	SAME	172	1	Mexican; fibroblast culture
AG3504		R1640	20I 0309	41	F	WH	+-(0)	MOTHER	172	2	Mexican; lymphoblast culture
AG3512	2	HF12	20U 0309	41	F	WH	+-(0)	MOTHER	172	2	Mexican; fibroblast culture
AG3505		R1640	20I 0309	37	M	WH	+-(0)	FATHER	172	3	Mexican; lymphoblast culture
AG3511	5	HF12	20U 0309	37	M	WH	+-(0)	FATHER	172	3	Mexican; fibroblast culture
AG3507		R1640	20I 0309	15	F	WH	+?	SIS	172	4	Mexican; lymphoblast culture
AG3508		R1640	20I 0309	17	M	WH	+?	BRO	172	5	Mexican; lymphoblast culture
AG3345		R1640	20I 0309	35	F	WH	+-(0)	MOTHER	543	1	French-Canadian; lymphoid culture
AG3346		R1640	20I 0309	35	M	WH	+-(0)	FATHER	543	2	French-Canadian; lymphoid culture
*AG3344		R1640	20I 0309	10	M	WH	--	PROBAND	543	3	Classical phenotype; lymphoid culture
AG3343		R1640	20I 0309	13	F	WH	+?	SIS	543	4	Clinically normal; lymphoid culture
AG3342		R1640	20I 0309	11	M	WH	+?	BRO	543	5	Minimal mental retardation; lymphoid culture
AG3198	3	HF12	20U 0309	10	F	WH	--	PROBAND	580	1	Mild maturity onset diabetes; skin fibroblast culture (dorsal forearm)
AG3198A	8	MC5A	20U 0309	10	F	WH	--	SAME	580	1	Mild maturity onset diabetes; skin fibroblast culture (leg)
AG3199	2	HF12	20U 0309	10	F	WH	--	SAME	580	1	Mild maturity onset diabetes; skin fibroblast culture (lateral hip)
AG3259A		R1640	20I 0309	10	F	WH	--	SAME	580	1	Same patient as AG3198; mild maturity onset diabetes; lymphoid culture
AG3257		MC5A	20U 0309	35	F	WH	+-(0)	MOTHER	580	2	Skin fibroblast culture; 4 passages at IMR
AG3260A		R1640	20I 0309	35	F	WH	+-(0)	MOTHER	580	2	Lymphoid culture

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
HUMAN CELL CULTURES: AGING											
Progeria (See GM Repository) - 26410											
AG3258		HF12 20U	0309	36	M	WH	+-(O)	FATHER	580	3	Fibroblast culture; 5 passages at IMR
AG3262A		R1640 20I	0309	6	F	WH	+?	SISTER	580	4	Lymphoid culture
AG3263A		R1640 20I	0309	12	M	WH	+?	BROTHER	580	5	Lymphoid culture
Progressive Supranuclear Palsy											
AG3674		R1640 15I	0348	60	M	WH		PROBAND	530	1	Bradykinesia; Parkinson speech; slight rigidity; no tremor; lymphoid culture
AG3675	4	HF12 20U	0348	60	M	WH		SAME	530	1	Bradykinesia; Parkinson speech; slight rigidity; no tremor; fibroblast culture
Rothmund-Thomson Syndrome - 26840											
AG3587	3	HF12 20U	0295	7	M	WH	--	PROBAND	545	1	Short stature; pancreatic insufficiency; normal intelligence; fibroblast culture
Spinal Muscular Atrophy											
AG3005	2	EEMEM 20U	0348	50	M	WH		PROBAND			Skin;progressive muscular weakness and wasting(proximal and distal);evidence of denervation;no sensory abnormality;no other family members affected
AG3730	3	HF12 20U	0348	54	M	WH		PROBAND	528	1	Polio at age 12; two cousins affected with similar spinal muscular atrophy; possibly is a late progression of polio; fibroblastic culture from muscle biopsy
AG3848	2	HF12 20U	0348	6	M	WH		PROBAND	581	1	Skin fibroblast culture; clinical findings compatible with Werdnig-Hoffman Type II
AG3849	3	EEMEM 20U	0348	27	F	WH		MOTHER	581	2	Skin fibroblast culture; mother of AG3848
AG3927		R1640 20I	0348	27	F	WH		MOTHER	581	2	Lymphoblastoid culture
Spinocerebellar Ataxia											
AG3818	5	EEMEM 20U	0348	45	F	WH		PROBAND			Skin biopsy; spasticity, increased reflexes, periperal neuropathy, distal atrophy; also has diabetes
AG3819	4	EEMEM 20U	0348	29	F	WH		PROBAND	555	1	Skin biopsy; ataxia, distal weakness and atrophy, progressive external ophthalmoplegia
AG3820	3	EEMEM 20U	0348	49	F	WH		MOTHER	555	2	Skin biopsy; ataxia, distal weakness and atrophy, progressive external ophthalmoplegia

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
HUMAN CELL CULTURES: AGING											
Thanatophoric Dwarfism - 27365											
AG0711	2	MC5A	20U 0143	1DA	M	WH	--	PROBAND	182	1	Fibroblast culture; cartilage
AG0712	2	MC5A	20U 0143	1DA	M	WH	--	SAME	182	1	Fibroblast culture; spleen
AG0713	2	MC5A	20U 0143	1DA	M	WH	--	SAME	182	1	Fibroblast culture; lung
AG0714	3	MC5A	20U 0143	1DA	M	WH	--	SAME	182	1	Fibroblast culture; skin
Translocation											
AG1839	7	HF12	20U 0031	10	F	WH		PROBAND	183	1	Hemihypertrophy; 45,XX,-13,-7,+t(7;13); fibroblast culture from skin of left arm
AG1840	7	HF12	20U 0031	10	F	WH		SAME	183	1	Left side of body is hypertrophied; 45,XX,-13,-7,+t(7;13);fibroblast culture from skin of right arm
Werner Syndrome - 27770											
AG0780	5	EEMEM	20U 0126	54	M	WH	--	PROBAND			Skin fibroblast culture
AG3141	1	HF12	20U 0319	30	F	WH	--	PROBAND			Skin;short stature;cataract;pigmented & atrophic skin;Hyperlipidemia type V;random chromosome rearrangements in 5/6 cells examined; tentative diag.:Werner
AG3829		R1640	20I 0309	42	M	AI	--	PROBAND	541	1	Lymphoblastoid culture; HLA type A:2, A:24,B:51,BW:35,CW:4
AG3593		EEMEM	20U 0044	28	M	WH	--	PROBAND	546	1	Fibroblast culture; 46,XY,t(5;8)(q11; q22),der dup(6)(p21>p25); no increased chromosome breakage or sister chromatid exchange; passage 4 at IMR
AG3594		EEMEM	20U 0044	30	M	WH	--	AFF BRO	546	2	Fibroblast cult.;46,XY,inv(3)t(3;13)(3qter>3q29::3q23>3q29::3q23>3p21::13q32>13qter;13pter>13q32::3p21>3pter),inv(9)(pter>p11::q34>p11::q34>qter)passage4IMR
AG3364A		R1640	20I 0309	42	M	WH	--	PROBAND	556	1	Typical phenotype; lymphoid culture
Xeroderma Pigmentosum: Complementation Group Undetermined - 27870											
AG1951	12	EEMEM	20U 0201	40	M	WH	--	PROBAND			From USSR; skin fibroblast culture; 46,XY,t(17;20)(17pter>17q25::20q13>20qter;20pter>20q13::17q25>17qter); LHC 638
AG1952	12	EEMEM	20U 0201	7	M	WH	--	PROBAND			From USSR; De Sanctis Cacchione syndrome; skin fibroblast culture; LHC 484

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
HUMAN CELL CULTURES: AGING											
Xeroderma Pigmentosum: Complementation Group Undetermined - 27870											
AG2592	3	HF12 20U	0226		M		--	PROBAND	310	1	Skin fibroblast culture; (possible XP?)
AG2593		R1640 20I	0226		M		--	SAME	310	1	Lymphoid cell culture; (possible XP?)
AG2611	3	HF12 20U	0226		F		+-(0)	MOTHER	310	2	Skin fibroblast culture
AG2872	3	HF12 20U	0023	24	F	WH	--	PROBAND	318	1	Skin fibroblast culture; XP1SA; Chilean
AG2871	3	HF12 20U	0023	25	F	WH	+?	SIS	318	2	Skin fibroblast culture; clinically unaffected; possible heterozygote
TUMOR PATIENTS											
Acute Myelogenous Leukemia											
*AG2655	2	HF12 20U	0195	26	F	WH		PROBAND	308	1	Affected; skin fibroblast culture; 15 years after treatment; 46,XX
*AG2648		R1640 20U	0195	18	M	WH		BRO	308	2	Unaffected; twin of AG2646; lymphoid culture
*AG2649	2	HF12 20U	0195	18	M	WH		BRO	308	2	Skin fibroblast culture; 46,XY
*AG2646		R1640 20U	0195	18	M	WH		BRO	308	3	Unaffected; twin of AG2648; lymphoid culture
*AG2647	2	HF12 20U	0195	18	M	WH		BRO	308	3	Skin fibroblast culture; 46,XY
*AG2653	3	HF12 20U	0195	42	F	WH		MOTHER	308	4	Skin fibroblast culture; treated with radium for cervical cancer 15 years earlier at the age of 27; 46,XX
*AG3607	2	HF12 20U	0195	44	F	WH		MOTHER	308	4	Skin; same as AG2653, 2 years later; rectal cancer; 46,XX/46,XX,t(12;13)(12qter>12p11::13q34>13qter;13pter>13q34::12p11>12pter),56%/44%
*AG2650		R1640 20U	0195	46	M	WH		FATHER	308	5	Unaffected; lymphoid culture
*AG2651	3	HF12 20U	0195	46	M	WH		FATHER	308	5	Skin fibroblast culture; 46,XY,t(5;8)(5qter>5p11::8q11>8qter;8pter>8q11::5p11>5pter)
Basal Cell Nevus Syndrome - 10940											
*AG3499	2	HF12 20U	0195	11	F	WH	+-	PROBAND			Skin; 46,XX/46,XX,t(5;X)(q35;q11),(85%/15%);treated with radio-& chemo-therapy; mother has Charcot-Marie-Tooth disease and naevoid basal cell carcinoma
Cancer Family											
AG3307	3	HF12 20U	0312	13	F	WH		AFF SIS	519	1	Skin fibroblast culture; benign junctional nevus

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
TUMOR PATIENTS											
Cancer Family											
AG3308	2	HF12	20U 0312	17	M	WH		AFF BRO	519	2	Skin fibroblast culture; liposarcoma of scrotum; oral papilloma; achondroplasia
AG3309	5	HF12	20U 0312	21	F	WH		SIS	519	3	Clinically unaffected; fibroblast culture
AG3310	2	HF12	20U 0312	46	F	WH		MOTHER	519	4	Neurofibroma; skin fibroblast culture
AG3311	4	HF12	20U 0312	51	M	WH		FATHER	519	5	History of cysts; fibroblast culture
AG3302	1	HF12	20U 0312	15	M	WH		PROBAND	519	6	Skin fibroblast culture; two primary malignancies:osteogenic sarcoma(metastatic) & neurogenic tumor(ganglion); positive family history of cancer
Chronic Myelogenous Leukemia											
AG1731	6	HF12	20U 0094	19	F			PROBAND	184	1	Irradiated skin
AG1732	3	HF12	20U 0094	19	F			SAME	184	1	Non-irradiated skin; lymphocytes show the Philadelphia marker chromosome; patient previously had Wilm's Tumor
Glomus Tumor - 13800											
AG0716	3	MC5A	20U 0159		F			PROBAND			Fibroblast culture from tumor
Medulloblastoma - 10940											
AG3000	3	HF12	20U 0195	7	M	WH		PROBAND	321	1	Skin fibroblast culture; brain tumor treated with radiation; Hodgkins disease ; 46,XY; 25% of cells showed evidence of chromsome and chromatid breakage
AG3001	4	HF12	20U 0195	41	F	WH		MOTHER	321	2	Unaffected; skin fibroblast culture; 46,XX
AG3002	5	HF12	20U 0195	19	M	WH		BRO	321	3	Unaffected; skin fibroblast culture; father had medulloblastoma;46,XY/47,XY, +18; (50%/42%)
Multiple Myeloma - 25450											
AG1311		R1640	20I 0156	68	M	BL		PROBAND	185	1	Lymphoblastoid culture - bone marrow; HLA type A10,A11(A3),B12
AG1312		R1640	20I 0156	68	M	BL		SAME	185	1	Lymphoblastoid culture - blood; HLA type A10,A11(A3),B12
AG1360	3	MC5A	20U 0156	68	M	BL		SAME	185	1	Skin fibroblast culture; same patient as AG1485
AG1485	3	MC5A	20U 0156	68	M	BL		SAME	185	1	Skin fibroblast culture; See also AG1311 Bone marrow; AG1312 Lymphocyte; and AG1360 Skin

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
TUMOR PATIENTS											
Multiple Primary Carcinoma											
AG2360	9	EEMEM 10I	0195	74	F			PROBAND			Nine different primary cancers; skin fibroblast culture; 46,XX
Nephroblastoma											
AG2508	3	HF12 20U	0242	7	F			PROBAND			Skin fibroblast culture; 46,XX
AG2514	2	HF12 20U	0242	8MO	F	WH		PROBAND			Skin fibroblast culture; 46,XX
Neurofibromatosis (von Recklinghausen Disease) - 16220											
AG3515	2	HF12 20U	0338	4	F	WH	+-	PROBAND			Skin;also has xanthogranuloma and myelomonocytic leukemia; 46,XX; treated with chemotheraputic agents; father and 2 sibs have neurofibromatosis
AG3516	2	HF12 20U	0338	3	M	WH	+-	PROBAND			Fibroblast culture; also has xanthogranuloma and myelomonocytic leukemia; 46,XY;25% of the cells have inconsistent abnormalities
Neuroblastoma - 25670											
AG2272	3	HF12 20U	0094	1	M			PROBAND			Skin fibroblast culture
*AG3320	51	EEMEM 10I	0152	13MO	M	WH		PROBAND			IMR-32; ATCC CCL 127; fibroblast and neuroblast-like cell culture; G6PD type B
AG2202	3	HF12 20U	0094	9	F			PROBAND	186	1	Non-irradiated skin; 46,XX
AG2203	3	HF12 20U	0094	9	F			SAME	186	1	Irradiated skin;46,XD+ partial trisomy 1
Osteogenic Sarcoma - 25950											
AG2086	2	HF12 20U	0094	20	M			PROBAND			46,XY; 11% of cells show inconsistent abnormalities; skin fibroblast culture
AG2308	2	HF12 20U	0094	2	F			PROBAND	312	1	Irradiated skin; first diagnosed as neurofibroma; 46,XX, some breaks
AG2309	2	HF12 20U	0094	2	F			SAME	312	1	Non-irradiated skin; 46,XX, multiple breaks and rearrangements
Retinoblastoma, Hereditary - 18020											
AG1262	4	EEMEM 20U	0198	7WK	F	WH	+-	PROBAND			Bilateral; conjunctiva fibroblast culture
AG1978	4	EEMEM 10I	0198	2	M		+-	PROBAND			Bilateral; conjunctiva fibroblast culture; 46,XY/46,XY,t(2;15)(p11;q22), t(3;12)(q21;q22),t,(8;15)(p21;q26), 46%/54%

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
TUMOR PATIENTS											
Retinoblastoma, Hereditary - 18020											
AG3139	2	HF12	20U 0195	17	M	BL	+-	PROBAND			Skin fibroblast culture; bilateral retinoblastoma onset at age 2; osteosarcoma of femur; family hx includes half-brother with retinoblastoma;46,XY
*AG1879	2	HF12	20U 0155	11	F	WH	+-	PROBAND	118	1	Bilateral; also has osteosarcoma; 46,XX; fibroblast culture
*AG1880	3	HF12	20U 0155	37	F	WH	+-	MOTHER	118	2	Unilateral retinoblastoma; 46,XX; skin fibroblast culture
AG3313	3	HF12	20U 0155	40	M	WH	++	FATHER	118	3	Skin fibroblast culture; 46,XY/45,XO; (76%/24%)
*AG1123	3	MC5A	20U 0094	9MO	F	BL	+-	PROBAND	189	1	Bilateral;early onset;multifocal;identical twin of AG1131;no other family members affected;46,XX;conjunctiva fibroblast culture
*AG1131	3	EEMEM	20U 0094	9MO	F	BL	+-	IDENT TWIN	189	2	Bilateral;early onset;multifocal;identical twin of AG1123;no other family members affected;46,XX;conjunctiva fibroblast culture
AG3326	2	HF12	20U 0320	3	F	WH	+-	PROBAND	431	1	Bilateral; also pinealoblastoma; scalp fibroblast culture
AG3641		R1640	20 0320	4	F	WH	+-	SAME	431	1	Bilateral; also pinealoblastoma; lymphoid culture
AG3640		R1640	20I 0320	37	F	WH	+-	AFF MOTHER	431	2	Bilateral; lymphoid culture
AG3376	2	HF12	20U 0094	2	F	WH	+-	PROBAND	576	1	Skin fibroblast culture; father also has bilateral retinoblastoma
AG3664	4	EEMEM	20U 0094	2	F	WH	+-	SAME	576	1	Father also has bilateral retinoblastoma; fibroblast culture derived from tumor(lumbar spinal cord)
*AG1408	4	MC5A	20U 0065	8MO	F	WH	+-	PROBAND	999	1	Bilateral; left eye conjunctiva fibroblast culture
*AG1980	5	EEMEM	10I 0198	1	F	WH	+-	SAME	999	1	Right eye conjunctiva fibroblast culture
Retinoblastoma, Sporadic											
*AG1946	6	EEMEM	10U 0198	18MO	M		+?	PROBAND			Unilateral; conjunctiva fibroblast culture; 46,XY

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				TUMOR PATIENTS								
Retinoblastoma, Sporadic												
*AG1947	6	EEMEM 10U	0198	2	M		+?		PROBAND			Unilateral; conjunctiva fibroblast culture; 46,XY,16qh+
*AG1979	4	EEMEM 10U	0198	2	M		+?		PROBAND			Unilateral; conjunctiva fibroblast culture; 46,XY
*AG2718	7	EEMEM 15U	0244	7	F		+?	A	PROBAND			Del(13); fibroblast culture; 46,XX,del (13)(pter>q12::q14>qter)
AG2971	14	MC5A 20U	0240	5MO	F	WH	+?		PROBAND			Translocation (X;13), breakpoint in 13q14; fibroblast culture
AG0913	3	EEMEM 20U	0065	2	M	WH	+?		PROBAND	117	1	Unilateral; fibroblast culture derived from tumor tissue
AG0914	3	EEMEM 20U	0065	2	M	WH	+?		SAME	117	1	Unilateral, conjunctiva fibroblast culture, 46,XY,9qh+
*AG1142	4	EEMEM 20U	0094	27MO	F	WH	+?		PROBAND	187	1	Bilateral; 46,XX,del(13)(q14q22); skin fibroblast culture; see also AG3022
AG1484		R1640 20I	0094	27MO	F	WH	+?		SAME	187	1	Lymphoblastoid culture
AG1231	3	EEMEM 20U	0065	22MO	F	WH	+?		PROBAND	188	1	Unilateral; conjunctiva fibroblast culture
*AG1232	3	R1640 20I	0065	22MO	F	WH	+?		SAME	188	1	Tumor tissue culture, grows in suspension
Sarcoma												
AG1517	3	EEMEM 20U	0094	9	F				PROBAND			Undifferentiated; skin fibroblast culture
Wilms Tumor - 27780												
AG2089	6	HF12 20U	0094	19	F		--		PROBAND			Fibroblast culture of irradiated skin
AG2510	2	HF12 20U	0242	3	F		--		PROBAND			Skin fibroblast culture; 46,XX
AG3485		R1640 20I	0195	13	F	WH	--		PROBAND			Lymphoid culture; chromosomes #11 appear normal in peripheral leukocytes; five family members with Wilm's tumor
AG3690	2	EEMEM 20U	0094	11MO	F	BL	--		PROBAND			Hemihypertrophy;gigantism;tumor biopsy; fibroblast culture
*AG1615	3	HF12 20U	0160	9	M	BL	--		PROBAND	190	1	Non-irradiated skin; fibroblast culture
AG1616	3	HF12 20U	0160	9	M	BL	--		SAME	190	1	Irradiated skin; fibroblast culture

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
TUMOR PATIENTS											
Wilms Tumor - 27780											
AG2129	2	HF12	20U 0199	10	M	BL	--	PROBAND	191	1	Fibroblast culture from non-irradiated skin
AG2130		R1640	20I 0199	10	M	BL	--	SAME	191	1	Lymphoblastoid cell culture
AG1894	2	HF12	20U 0094	12	M	WH	--	PROBAND	192	1	Irradiated skin, fibroblast culture
AG1895	2	HF12	20U 0094	12	M	WH	--	SAME	192	1	Non-irradiated skin, fibroblast culture
AG2511	2	HF12	20U 0242	5MO	M		--	PROBAND	314	1	Fibroblast culture derived from tumor tissue; 46,XY
AG2512	2	HF12	20U 0242	5MO	M		--	SAME	314	1	Skin fibroblast culture; 46,XY
AG2310	3	HF12	20U 0094	3	M		--	PROBAND	346	1	Irradiated skin; 46,XY, abnormal chromosomes:40%; 46,XY,-22,+t(17p+;22q-) +fragment,40%; multiple breaks and rearrangements
AG2311	2	HF12	20U 0094	3	M		--	SAME	346	1	Non-irradiated skin; 46,XY; 9% of cells show abnormal chromosomes
APPARENTLY NORMAL HUMAN FIBROBLAST CELL CULTURES											
Apparently Normal: Non-Fetal Tissue											
AG1437	2	MC5A	20U 0152	3DA	M	WH		PROBAND			Foreskin
AG1439	3	MC5A	20U 0152	3DA	M	BL		PROBAND			Foreskin
AG1440	3	MC5A	20U 0152	3DA	M	BL		PROBAND			Foreskin
*AG1518	2	MC5A	20U 0152	3DA	M			PROBAND			46,XY; foreskin
AG1519	2	MC5A	20U 0152	3DA	M			PROBAND			46,XY; foreskin
AG1520	2	MC5A	20U 0152	3DA	M			PROBAND			Foreskin
AG1521	2	MC5A	20U 0152	3DA	M			PROBAND			Foreskin
*AG1522	2	MC5A	20U 0152	3DA	M			PROBAND			Foreskin
AG1523	2	MC5A	20U 0152	3DA	M			PROBAND			46,XY; foreskin
AG2101	8	MC5A	10U 0203	27	F			PROBAND			46,XX; uterus, endometrium
AG2102	4	EEMEM	20U 0203	27	F			PROBAND			46,XX; uterus, endometrium
AG2220	3	HF12	20U 0200	22	M	WH		PROBAND			Skin

AGING CELL REPOSITORY CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
APPARENTLY NORMAL HUMAN FIBROBLAST CELL CULTURES											
Apparently Normal: Non-Fetal Tissue											
*AG2222	2	HF12	20U 0200	49	M	WH		PROBAND			Skin
*AG2257	1	HF12	20U 0200	46	F	WH		PROBAND	196	1	Skin
*AG2258	4	HF12	20U 0200	46	F	WH		SAME	196	1	Lung
AG2261	2	HF12	20U 0200	61	M	WH		PROBAND	311	1	Skin
AG2262	1	HF12	20U 0200	61	M	WH		SAME	311	1	Lung
AG2602	3	HF12	20U 0200	35	M			PROBAND	316	1	Skin
AG2603	2	HF12	20U 0200	35	M			SAME	316	1	Lung
SV40 VIRUS TRANSFORMED CELL CULTURES											
IMR90											
AG2804A	41	MC5A	20U 0135	16FW	F	WH		PROBAND			SV40 transformed IMR90; post-crisis, T-antigen positive, 1-5% V-antigen positive
Retinoblastoma, Sporadic											
AG3022	49	HF12	20U 0135	27MO	F	WH		PROBAND			SV40 transformed AG1142; 100% T-antigen pos.,1-5% V-antigen pos.,post-crisis line near tetraploid;mult.rearrangement; deleted chrom. 13 present in 90% cells
SPECIALLY CHARACTERIZED DIPLOID CELL CULTURES											
Large Quantity of Characterized Cells Frozen at Early Passage											
*IMR90	5	MC5A	20U 0026	16FW	F	WH	A	PROBAND			Lung; see guidelines published in Science 196:60-63 (1977); available at population doubling levels: 12.3,21,25, 27,30,and 32
*IMR91	5	MC5A	20U 0026	14FW	M	WH	A	PROBAND			Lung; available at population doubling levels: 15,19,22,26,29,32,and 37

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APPENDIX A

CODE FOR CULTURE MEDIA

For reference see: Morton, Helen C., A survey of Commercially Available Tissue Culture Media. In Vitro 6:89-108, 1970.

The code consists of an abbreviation of the culture medium followed by a number, indicating the percentage of fetal calf serum, and the letter I or U. I indicates that the fetal calf serum is heat inactivated at 60°C for 1/2 hour before it is added to the culture medium. U indicates that the fetal calf serum is used without prior inactivation (uninactivated). The abbreviations are as follows:

EEMEM - Minimum Essential Medium Eagle in Earle BSS with 2x concentration of essential and non-essential amino acids and vitamins.

HEMEM - Minimum Essential Medium in Hank BSS, with 2x concentration of essential and nonessential amino acids and vitamins.

R1640 - Roswell Park Memorial Institute Medium 1640

Mc5A - McCoy 5A Medium, Iwakata and Grace Modification

DUMEM - Dulbecco Minimal Essential Medium

HF12 - Ham's F12 Medium

HF10 - Ham's F10 Medium

APPENDIX B

SUBMITTER CODE

THE HUMAN GENETIC MUTANT CELL REPOSITORY IS INDEBTED TO THE FOLLOWING INVESTIGATORS WHO GRACIOUSLY SUBMITTED BIOPSIES, CULTURES, AND DATA FOR CHARACTERIZATION.

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0002 Alfi	O	Childrens Hosp of LA, Div Med Gen 4650 Sunset Blvd LosAngeles CA 90054 USA
0003 Allderdice	P	Memorial U Dept of Medicine St Johns NF Canada
0004 Aula	P	U of Helsinki Dept of Pathology Helsinki 00290 Finland
0005 Barranco	S	U of Texas Med Sch Div of Cell Biology Galveston TX 77550 USA
0006 Beaudet	A	Baylor Col Med, Dep of Ped Rm 325E 1200 Moursound Ave Houston TX 77025 USA
0007 Benke	P	U of Miami Sch Med, Mailman Cntr PO Box 520006 Miami FL 33152 USA
0008 Beratis	N	Med Gen Ins for Basic Res Men Ret 1050 Forest Hill Road Staten Isl NY 10314 USA
0009 Beutler	E	City of Hope Med Cen, Dept Med 1500 East Durate Rd Duarte CA 91010 USA
0010 Bloom	A	Columbia U Col Phys & Surg Dep Ped, 701 W 168th St New York NY 10032 USA
0011 Bootsma	D	Erasmus U Rotterdam Dep Cell Biol, Postbus 1738 Rotterdam Netherland
0012 Borgaonkar	D	N Texas State U Genetics Center Denton TX 76203 USA

0013 Boue	A	Centre d'Etudes de Biologie Prentale Chateau de Longchamp, Bois de Boulogne Paris 75016 France
0014 Bowen	P	U of Alberta Faculty of Med Dept of Ped, 4-120 Clinical Sci Bldg Edmonton Al T6G 2E1 Canada
0015 Breg	W	Yale U School of Medicine Dep Hum Gen, 333 Cedar St New Haven CT 06510 USA
0016 Brown	J	Medical Col of Virginia Dep Human Genetics Richmond VA 23298 USA
0017 Buchanan	P	U North Carolina Sch Med Dep Ped ChapelHill NC 27514 USA
0018 Bucknall	W	Baylor Col of Med Med Gen Sec, Dep of Med Houston TX 77025 USA
0019 Buist	N	U of Oregon, Health Sci Cen, Dep Ped 3181 Sam Jackson Park Rd Portland OR 97201 USA
0020 Bull	M	New England Med Cen Dep of Ped Boston MA 02111 USA
0021 Chen	H	Children's Hosp of Michigan Beaubien Blvd Detroit MI 48201 USA
0022 Chu	E	U of Michigan Med Sch, Dep Human Gen 1137 E Catherine St Ann Arbor MI 48104 USA
0023 Cleaver	J	U Calif Med Sch at San Francisco Lab of Radiobiology San Fran CA 94143 USA
0024 Conod	E	Albert Einstein Col Med, Pediatrics 1300 Morris Park Ave Bronx NY 10461 USA
0025 Conover	J	Albert Einstein Col of Med, Pediatrics 1300 Morris Park Ave Bronx NY 10461 USA
0026 Coriell	L	Institute for Medical Research Copewood St Camden NJ 08103 USA

0027 Cox	R	New York U Med Cntr, Sch Med 550 First Ave New York NY 10016 USA
0028 Croce	C	Wistar Institute 36th and Spruce Sts. Phila PA 19104 USA
0029 Davignon	J	Clinical Research Inst 110 Pine Ave, West Montreal Qu H2W 1R7 Canada
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0031 DeMars	R	U of Wisconsin Dep of Genetics Madison WI 53706 USA
0032 Desnick	R	Mt Sinai Med Sch, Peds, Div Med Gen Fifth Ave & 100 St New York NY 10029 USA
0033 Elsas	L	Emery U Sch Med Div Med Genetics Atlanta GA 30322 USA
0034 Fajans	S	U Michigan Med Cen Metabolism Research Unit Ann Arbor MI 48104 USA
0035 Fand	I	SUNY at Stony Brook Dep Psych, Heal Sci Cen Tower Stonybrook NY 11794 USA
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0037 Fialkow	P	U Washington Sch Med Div Med Gen Seattle WA 98195 USA
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U California Sch of Med
Dep Obs & Gyne
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Tagensivej-Copenhagen N
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Dep Cancer Studies
Birmingham B15 2TJ England

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0071 Kelley	W	U Michigan Med Cen Dep Int Med Ann Arbor MI 48109 USA
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0073 Kim	H	Beth Israel Med Cen, Dep Ped 421 East 14th St New York NY 10009 USA
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0075 Klinger	H	Albert Einstein Col, Dep Gen 1300 Morris Park Ave New York NY 10461 USA
0076 Knittle	J	Mt Sinai Sch Med, Dep Ped 5th Ave and 100th St New York NY 10029 USA
0077 Kohn	G	Haddassah-Hebrew U Med Center Dept Human Genet, P O Box 499 Jerusalem Israel
0078 Kousseff	B	S Illinois U Sch Med, Dep Ped PO Box 3926 Spgfield IL 62708 USA
0079 Krompotic	E	Mt Sinai Hosp Med Cen Dep Exp Biology Chicago IL 60608 USA
0080 Krooth	R	Columbia U, Hlth Sci Bldg Rm 1311 710 W 168th St New York NY 10032 USA
0081 Kushnick	T	N J Col Med, Dep Ped 100 Bergen St Newark NJ 07103 USA
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0083 Littlefield	J	Johns Hopkins U Sch Med Dep Ped Baltimore MD 21205 USA
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0088 Mahoney	M	Yale U Sch Med, Dep Hum Gen 333 Cedar St New Haven CT 06510 USA
0089 Manduka	M	Institute for Medical Research Copewood St Camden NJ 08103 USA
0090 Marks	J	U Texas Heal Sci Cen at Dallas 5323 Harry Hines Blvd Dallas TX 75235 USA
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0092 Maumenee	I	The Wilmer Opthalmologic Inst 601 N Broadway Baltimore MD 21205 USA
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0094 Meadows	A	Children's Hosp of Phila 34th and Civic Cntr Blvd Phila PA 19104 USA
0095 Mellman	W	U Pennsylvania Sch Med, Dep Hum Gen G/4 Richards Bldg Phila PA 19174 USA
0096 Migeon	B	Johns Hopkins U Sch Med, Dep Ped 601 N Broadway Baltimore MD 21205 USA

0097 Mao	T S	Training and Research Center Bordentown NJ 08505 USA
0098 Miwa	S	Yamaguchi U of Med Third Dep Internal Med Yamaguchi 755 Japan
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0100 Moshug	T	Hahnemann Med Col, Dep Ped 230 N Broad St Phila PA 19102 USA
0101 Nyhan	W	U Calif at San Diego Sch Med, Dep Ped PO Box 109 La Jolla CA 92037 USA
0102 Orkin	S	Childrens Hosp Med Cen, Dep Med 300 Longwood Ave Boston MA 02115 USA
0103 Parkman	R	Childrens Hosp Med Cen, Lab of Immun 300 Longwood Ave Boston MA 02115 USA
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0105 Phillippart	M	U Calif, Neuro-Psych Inst, Rm 48-228-B 760 Westwood Plaza LosAngeles CA 90024 USA
0106 Pope	I	Madigan Army Med Cen Dep Ped Tacoma WA 98431 USA
0107 Punnett	H	St Christophers Hosp, Dep Cytogen 2600 N Lawrence St Phila PA 19133 USA
0108 Romeo	G	Stanford Med Sch Dep Human Genetics Stanford CA 94305 USA
0109 Rary	J	Johns Hopkins U Sch Med Rm 203 Biophysics Bldg 4 Baltmore MD 21205 USA
0110 Robbins	J	NIH, National Cancer Inst Bldg 10, Rm 12N 258 Bethesda MD 20205 USA

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0114 Schneider	J	U Calf at San Diego, Dep Ped PO Box 109 La Jolla CA 92037 USA
0115 Schmickle	R	Univ Hosp, Dep Ped 2015 Holden Perinatal Res Blvd Ann Arbor MI 48104 USA
0116 Schuh	B	Woodbridge State School Woodbridge NJ 07905 USA
0117 Schulman	J	NIH Bldg 10, Rm 13N-256 Bethesda MD 20205 USA
0118 Scott	C	U Washington Sch Med Dep Ped Seattle WA 98195 USA
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0121 Segal	S	Childrens Hosp, Dep Ped & Med 34th & Civic Center Blvd Phila PA 19104 USA
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0127 Soter	N	Robert B Brigham Hosp 125 Parker Hill Ave Boston MA 02120 USA
0128 States	B	Children's Hosp of Philadelphia 34th and Civic Cntr Blvd Phila PA 19104 USA
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0131 Summer	G	U North Carolina Sch Med Dep Biochem ChapelHill NC 27514 USA
0132 Summitt	R	U Tennessee, Dep Ped, Genetics Sec 860 Madison Ave Memphis TN 38163 USA
0133 Swift	M	U North Carolina Child Dev Inst, Dep Med ChapelHill NC 27514 USA
0134 Thomas	G	John Kennedy Inst 707 N Broadway Baltimore MD 21205 USA
0135 Toji	L	Institute for Medical Research Copewood St Camden NJ 08103 USA
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0156 Vasso	S	Our Lady of Lourdes Hosp, Dep Pathology Haddon Ave Camden NJ 08103 USA
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0159 Murphy	D	National Inst of Aging, Pub Heal Serv Landow Bldg C-703 Bethesda MD 20205 USA
0160 D'Angio	G	Children's Hosp of Philadelphia 34th and Civic Cntr Blvd Phila PA 19104 USA
0161 Diamond	L	Wistar Institute 36th and Spruce Sts Phila PA 19104 USA
0162 Ampola	M	New England Med Cntr Hosp 171 Harrison Ave Boston MA 02111 USA
0163 Levine	E	Wistar Institute 36th & Spruce Sts Phila PA 19104 USA
0164 Carpenter	D	Sunland Hospital P O Box 3513 Orlando FL 32802 USA
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0166 Clark	W	Hartford Hosp Dep Pathology Hartford CT 06115 USA

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Rm L-5085 Sch of Med
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0179 Wasdahl W
U North Dakota
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GrandForks ND 58202 USA

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Cornell U Sch Med, Dep Radiology
525 E 68th St
New York NY 10021 USA

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U British Columbia
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0198 Felberg	N	Wills Eye Hosp, Dep Biochem 1605 Brandywine St Phila PA 19130 USA
0199 Lazarus	G	Duke U Med Cen Div Dermatology Durham NC 27710 USA
0200 Read	W	110 Munn Lane CherryHill NJ 08034 USA
0201 Boschkow	N	Cancer Research Cen, Med Genetics 1154783 Kashivskaye, Chaussee Moscow USSR
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0205 Moore	G	Denver General Hospital W 8th Ave & Cherokee St Denver CO 80204 USA
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0208 Hirschhorn	R	NYU Med Cen, Sch Med, Dep Med 550 First Ave New York NY 10016 USA
0209 Meisner	L	U Wisconsin, State Lab Hygiene 465 Henry Mall Madison WI 53706 USA

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0212 Jesseph	J	Indiana U Sch Med, Dep Surgery 1100 W Michigan St Indianapls IN 46202 USA
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0214 Aylsworth	A	U North Carolina Dep Peds ChapelHill NC 27514 USA
0215 Howard	B	NIAMDD, Phoenix Clinical Res Cen 4212 North 16th St, Rm 541 Phoenix AZ 85016 USA
0216 Zurier	R	U Connecticut Health Cen Dep of Med Farmington CT 06032 USA
0217 Anderson	J	Henry Ford Hosp, Div Allergy & Clin Imm W Grand Blvd Detroit MI 48202 USA
0218 Rosenberg	L	Yale U Sch Med, Dep Human Gen 904 LCI New Haven CT 06510 USA
0219 Polmar	S	Case Western Res Med Sch, Dep Peds Rainbow Lab & Child Hosp, 2103 Adelbert Cleveland OH USA
0221 Serra	A	U Catolica, Ins Genetia Umana Viadella Pineta Sacchetti 644 Rome 00168 Italy
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0223 Hong	R	U Wisconsin, Cen Health Science 1300 University Ave Madison WI 53706 USA
0224 Van Dyke	D	Henry Ford Hosp 2799 W Grand Blvd Detroit MI 48202 USA

0225 Krush	A	Johns Hopkins Hosp, The Moore Clinic Div Med Genetics Baltimore MD 21205 USA
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0227 Levis	W	NCI, NIH, Div of Dermatology Bldg 10, Rm 12N260 Bethesda MD 20205 USA
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0229 Mailhes	J	Louisiana State U Med Cen, Dep Peds Birth Defects Center Shreveport LA 71130 USA
0230 Cantz	M	Westfallsche Wilhelms Universitat Physiol Chem institut Munster 4400 W Germany
0231 Waterson	J	University Hosp, Dep Peds 2015 Holden Perinatal Res Bldg Ann Arbor MI 48104 USA
0232 McGuire	J	Yale U, Dep Dermatology 333 Cedar Ave New Haven CT 06510 USA
0234 Tashian	R	U Michigan, Dep Human Genetics 1137 E Catherine St Ann Arbor MI 48109 USA
0235 Frenkel	E	U Texas Health Science Center Dep Int Med Dallas TX 75235 USA
0236 Rubinstein	A	A Einstein Coll of Med 1300 Morris Park Ave Bronx NY 10461 USA
0237 Griscelli	C	Centre d'Etudes de Biologie Prenatale Chateau de Longchamp, Bois de Boulogne Paris 75016 France
0238 Ropers	H	Albert-Ludwig U, Ins Humangenetik Freibert I Briden Freiberg W. Germany
0239 Baserga	R	Temple U Health Science Center Dep Pathology Phila PA 19140 USA

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0242 Pogosyanz	H	Cancer Research Cen, Lab Cytogenetics Kashirokoje Shousse 6 Moscow M-478 USSR
0243 Farber	P	Geisinger Medical Center Cytogenetics Lab Danville PA 17821 USA
0244 Weichselbaum	R	Harvard U Sch Pub Health, Dep Physiology 665 Huntington Ave Boston MA 02115 USA
0245 Chatterjee	S	Johns Hopkins Hosp, Dep Ped Child Med Surg Cen, Rm 1102 Baltimore MD 21205 USA
0246 Pyatt	E	St Barnabas Med Cen, Cytogenetics Lab Old Short Hills Rd Livingston NJ 07039 USA
0247 Cowell	H	A I duPont Institute, Clin Res Dep PO Box 269 Wilmington DE 19899 USA
0248 Lutzner	M	NIH, NCI, Dermatology Branch BLdg 10 Rm 12N238 Bethesda MD 20205 USA
0249 Brauermann	R	Yale U New Haven CT 06510 USA
0250 Gedd-Dahl	D	Norsk Hydro Ins for Cancer Research Genetics Lab, Montebello Oslo 3 Norway
0251 Meuwissen	H J	Union U Med Col Dep Pediatrics Albany NY 12208 USA
0252 McCaw	B	Southwestern Biomed Res Inst P O Box 1031, 123 University Dr Tempe AR 85281 USA
0253 Desposito		New Jersey Col Med Dep Ob & Gyn Newark NJ 07103 USA

0255 Salazar	E	Catedra de Fisiopatologia ULA Laboratorio de Homonas HULA Merida Venezuela
0256 Abel	A	3015 Stadium Dr Apt 2 Columbus OH 43202 USA
0257 Wohttmann	H	U South Carolina Sch Med 80 Barre St Charleston SC 29401 USA
0258 Kennedy	C	St George Hosp Dep Dermatology London England
0259 Goodenberger	D	NIH NCI Bethesda MD 20205 USA
0260 Buck	D	U Pa Sch Med, Dep Human Genetics Richards Bldg G/4 Phila PA 19174 USA
0262 Puck	T	U Colorado Sch Med Eleanor Roosevelt Ins Cancer Res Denver CO 80220 USA
0263 Haseltine	F	Yale U Sch Med Dep Ob & Gyn New Haven CT 06510 USA
0264 Hoar	D I	U Toronto Dep Med Genetics Toronto On Canada
0265 Palmer	G G	U Indiana Sch Med Dep Med Genetics Indianapls IN 46202 USA
0268 Friedrich	U	U Aarhus, Ins Human Genetics 8000 Aarhus C Aarhus Denmark
0269 Coldwell	J	Children's Medical Center 5330 E S Kelly St Tulsa OK 74135 USA
0270 Scott	C	U Texas Med Sch, Dep Peds John Freeman Bldg Houston TX 77025 USA
0272 Whitaker	J	1710 St Julian Place Columbia SC 29206 USA

0273 Gregson	N M	General Hosp, Wessex Reg Cytogen Unit Pathology Dep Wilts SP2 7SX U.K.
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APPENDIX C

References

The following pages list references which include clinical descriptions of patients and their families from whom specimens stored in the Repository were obtained as well as publications detailing investigations utilizing cultures obtained from the Repository. We hope to update this list annually to provide a comprehensive bibliography for each culture. This goal could be realized more easily if investigators would identify cultures in their publications by their GM or AG numbers and forward reprints to the Repository.

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APPENDIX D

NSF MASS CELL CULTURE CENTERS

The Mass Cell Culture Centers at the Massachusetts Institute of Technology and the University of Alabama at Birmingham are accepting applications for large scale cell and virus production required for highly meritorius research projects. These centers established by the National Science Foundation are intended to serve as a facility and research resource for scientists throughout the country.

The MIT Center is headed by Dr. Phillips W. Robbins of MIT and Dr. Richard L. Davidson of Harvard Medical School. This facility is designed for large scale monolayer and suspension cultures and has experience with a large number of different cell lines and viruses and also with diploid human fibroblasts. The University of Alabama Center is headed by J. Claude Bennett of the Department of Microbiology and Ronald T. Acton of the Diabetes Research and Training Center. This facility is designed for large scale suspension cultures with a capability of producing one kilogram or more of cells per week. Their experience to date is primarily with lymphoblastoid cell lines. The purpose of these centers is to produce cells and viruses on a large scale in order to allow scientists to conduct novel and important experiments in basic biological research that could not be accomplished with the materials and resources in the investigators' own laboratory. Approval of applications is on a competitive basis, based on the merit of the research project as evaluated by Centers' Steering Committees.

Examples of recent activities at the Centers are:

1. Production of 100 mg Sindbis Virus propagated in 200 roller bottles of cells for use in x-ray structural studies.

2. Production of 750 roller bottles of SV40 transformed BALB 3T3 cells for purification and characterization of T antigen.

3. Growth on 300 liters of mouse leukemia cells for isolation and structural analysis of a specific lysine transfer RNA implicated in the control of cell division.

4. Growth of 3.1×10^{12} BW 5147 cells for the isolation and structural analysis of H-2k and Thy-1 alloantigens.

Following approval of an application, the investigator sends a stock of cells or virus to the center. The stock is then grown to the requested amount, under conditions of careful handling and rigorous screening for contaminants (including mycoplasma), and prepared according to the needs of the investigator. Investigators will be asked to pay minimal costs to cover media and other supplies.

The procedure for applying involves submitting a letter specifying the amount of cell or virus required along with a brief description of the relevant research project and available supporting material. Applications or inquiries should be addressed to:

Mr. Donald Giard
Cell Culture Center
E17-321
Massachusetts Institute of Technology
77 Massachusetts Avenue
Cambridge, MA 02139

OR

Dr. Ronald T. Acton
Diabetes Research and Education Hospital
University of Alabama Hospitals and Clinics
1808 7th Avenue South
Birmingham, AL 35294

APPENDIX E

The following is a listing of Repository numbers versus page numbers. The GM and AG designations have been omitted for convenience. Following each culture number is the page(s) on which that cell culture may be found. The letters preceding the page numbers indicate the section of the catalog in which that page/culture is located: F, fibroblast; SV, virus transformed fibroblast; L, lymphocyte; A, amniotic; AN, animal; and AG, aging.

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